



HEREDITARY
NEUROPATHY
FOUNDATION

SPRING 2021

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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.

www.hnf-cure.org

CMT UPDATE SPRING 2021



Allison T. Moore

Founder and CEO

Hereditary Neuropathy Foundation

Dear Friends:

This summer marks seven years since the announcement that HNF had entered into a partnership with the French biopharmaceutical company, Pharnext, to help raise awareness of Charcot-Marie-Tooth (CMT) disease and support the CMT community through several initiatives. Pharnext's support has assisted HNF in distributing the CMT Update quarterly newsletter, enhanced the Global Registry for Inherited Neuropathies (GRIN), set-up activities for CMT September Awareness Month in the US and strengthened the CMT Inspire Online Community. Additionally, Pharnext has supported HNF's Patient-Centered Summits, including sponsorship of the Externally-led Patient-Focused Drug Development Meeting for the FDA. So it is with great enthusiasm, pride and support that we are announcing the recruitment phase of the Pharnext PREMIER Trial of PXT3003 – the first ever therapeutic treatment for CMT (specifically, CMT1A) – and the recent exciting results of the on-going study that show sustained treatment benefits for CMT1A patients treated with PXT3003 at High Dose. I hope that you will read all about it on page 2 of this newsletter and consider participating in the trial at a center near you.

In this post-pandemic world, I am excited to share that HNF has stepped up and continued to support the patient community with a robust offering of Movement is Medicine™ adaptive exercise classes on-line, as well as a virtual MiM™ Summit in partnership with AdventHealth. Not only are we committed to increasing awareness of the importance of exercise for those living with CMT, but we're having a ton of fun doing it! Nearly 600 of you joined me for the impromptu dance party, adapted exercise and informative sessions on March 7 and it was exhilarating! The wonderful comments we received after the event were so rewarding (see a sampling of them on page 6).

The project I am most proud of is our visual CMT Medical Journal. We are testing it with a small group of participants currently, but soon it will be available for everyone. With the goal to improve the time to diagnosis and delivery of expert care to everyone – whether you are near a CMT Center of Excellence (page 19) or not – you'll be able to share pictures and videos of your daily activities and challenges of living with CMT with our CMT experts. I'd like to personally thank Addie and her mom and dad for working with us to create all of the tutorial videos included with the app (and Leslie Nelson, PT, PhD, at UT Southwestern for coaching Addie through it all!).

The HNF team has been busier than ever: we've added a few more MiM™ Ambassadors, created informational content for neurologists and health care providers to be published in the annual Rare Neurological Disease Report, added a new Center of Excellence and a new board member. Estela has created an awesome Wishlist of super-helpful tools and gadgets on amazon.com (please take a minute to choose HNF as your charity of choice at smile.amazon.com) and she wrote about why advocacy is so important to her. You'll definitely be motivated to [Get Involved](#) after reading her article!

I hope that you are as energized as we are to continue fighting the fight together to raise the funds needed to raise awareness, provide support and resources, and bring treatments across the finish line for everyone living with CMT!

Your support is critical to achieve our mission – please [DONATE TODAY!](#)

Best,

Allison T. Moore



PHARNEXT PREMIER TRIAL OF PXT3003 CMT1A CLINICAL TRIAL – NOW SCREENING FOR PARTICIPANTS

HNF is proud to have 19 of its CMT Centers of Excellence participate in the **PREMIER Trial**, which is being conducted in patients with mild-to-moderate CMT1A. The International multi-centered pivotal Phase III study will enroll up to 350 subjects ages 16-65 at 50 sites worldwide (**20 in the US, 5 in Canada and the rest in Europe and Israel**).

The **PREMIER Trial** is a randomized, double-blind, two-arm placebo-controlled study designed to confirm PXT3003 safety and efficacy in patients with CMT1A. Patients enrolled will be treated for 15 months.

As agreed with regulatory agencies, the primary efficacy endpoint is the Overall Neuropathy Limitation Score (ONLS) scale, which measures functional motor disability. The secondary endpoints will include the following outcome measures: 1) 10-Meter Walk Test (10mWT), 2) Quantified Muscular Testing (bilateral foot dorsiflexion dynamometry), 3) Patient Global Impression of Severity (PGI-S), 4) Patient Global Impression of Change (PGI-C), 5) Charcot-Marie-Tooth Neuropathy Score, version 2 (CMTNS-v2), and 6) Quantified Muscular Testing (hand grip). Safety and tolerability will be monitored throughout the study.

Patients with a confirmed genetic diagnosis of CMT1A can fill out an eligibility questionnaire at <https://www.premiercmt1a.com> to assess potential participation in the Premier study.

****Pharnext is also continuing to conduct its ongoing Phase III Extension Study, CLN-PXT3003-03, an open label study that has enrolled 187 patients out of the 323 with mild-to-moderate CMT1A patients that were enrolled in the first double-blind Phase III Trial (PLEO-CMT). As of today, 130 CMT1A patients are still being treated with high-dose PXT3003 and have been followed for more than 2 years in order to assess not only the safety and tolerability of PXT3003, but also its long-term efficacy. Pharnext recently released a statement reporting top-line interim data on the safety and efficacy of PXT-3003 which [you can read here](#).*

MORE INFORMATION about the PREMIER Trial can be found at:
<https://clinicaltrials.gov/ct2/show/NCT04762758>







New Study Measures Progression of CMT1A Nerve Impairment

CONTRIBUTOR: DAN KNAUSS

Researchers in Japan [recently published the results of a study](#) that measured how much motor nerve recruitment has diminished over time and relative to unaffected, healthy neurons.

In this study, nineteen people with CMT1A were tested with high-density surface-electromyography (surface-EMG) equipment. The surface-EMG was applied to the test subjects' thighs during isometric knee extensions. (In this exercise, the foreleg is raised and elevated parallel to the floor in a sitting position.) The test focused only on the largest and strongest part of the quadriceps muscle, which extends the knee to move the lower leg.

The firing rate of individual motor units in the test subjects' legs was tested twice with the second test, following a year after the first. They also did the same series of tests with a control group of people who do not have CMT1A. The resulting data showed that CMT1A patients had lower instantaneous firing rates (IFRs) than those in the control group. Their motor units' firing rate decreased over time without any outwardly visible changes.

Since there was a clear and consistent drop in IFR for the CMT patients after a year, surface-EMG could be a tool for assessing the progression of CMT 1A.

By comparing the average firing rate for the CMT 1A and non-CMT 1A groups, it was also possible to see how much CMT1A has degraded someone's motor nerves and muscles relative to a healthy baseline.

Getting Under the Hood: Your Muscles' "Motor Pool"

The motor unit is the essential contractile structure in the muscles: a motor neuron located in the spinal cord, its axon, and innervating muscle fibers. Even though CMT1A is primarily a demyelinating hereditary neuropathy, axonal damage occurs along with demyelination.

A single motor unit's firing rate is proportionate to the amount of contractile force required by that unit. Groups of MUs called

a "motor pool" fire together to activate the muscle with the most vigorous possible contraction. With CMT 1A impairing the firing rate, the muscle contraction will be weaker even if a large motor pool is recruited.

Specific Conclusions About CMT1A's Impact and Progression

The specific neuromuscular impact of CMT1A, according to this study, involves all levels of force generation. In this study, there was no decrease in maximum voluntary contraction (MVC), a standard for measuring muscle strength in people with neuromuscular diseases relative to normal rates.

Ramp-up contractions in the test subjects with CMT1A averaged 10.3 pulses per second (pps) compared to 12.2 pps in the control group without CMT 1A. Throughout sustained contractions, the CMT 1A group averaged 8.0 pps compared to the control group's 9.3 pps. The average firing rates of the CMT1A patients fell, during sustained contractions, from 8.06 to 7.52 pps, indicating fatigue.

Notably, [an earlier study in](#) CMT1A patients using surface-EMG showed lower motor cortex activity in the brain when test subjects were planning their leg lifts. This study concluded that the brain is very tied to fatigue in CMT1A, with higher-than-normal activity in the prefrontal cortex, which may be compensating for the low motor cortex activation reported in this study for the patients with CMT 1A. Awareness of the strain and conscious effort to operate muscles could be a cause and effect of fatigue.

Anyone with CMT knows it's tiring to consciously struggle through pain and fatigue to make their arm or leg muscles move. Still, these studies provide measures and tools for assessing the severity and progression of the disease.

Meet Addie “The Face of the CMT Medical Journal”



In the Fall CMTUpdate, the feature article announced that HNF, in partnership with CaptureProof, is bringing innovative and powerful computer learning capabilities to give you the power to capture and document your CMT story with photos and videos to improve the health care you receive. We’re calling the project, The CMT Medical Journal. When it came time to film the tutorial videos for the project, we knew we wanted Addie to be “The Face of the CMT Medical Journal”. With coaching from Leslie Nelson, PT, PhD, Board Certified Specialist in Orthopedic Physical Therapy and Assistant Professor at UT Southwestern School of Health Professions Department of Physical Therapy, Addie demonstrates each of the exercises in the protocol like a pro!

Addie is 11 years old and lives in Austin, Texas. When she was 4 years old her parents noticed that she was walking oddly and after many doctor visits and testing she was diagnosed at 6 years old with CMT1A. We had a candid conversation with Addie about life with CMT and here’s what she told us, in her own words:

“ Having a CMT diagnosis is really interesting and rewarding. I have the ability to expand other people’s knowledge about CMT. At times CMT is difficult for me, my mom often helps me tie my shoes as I get ready for school. My friends run ahead of me and typing on the computer is challenging. My teacher often will type my notes for me.

There are many activities that I participate in that are complementary to my CMT: dance, swimming and acting.

One thing I like least about CMT is the odd looks people give me when I use the elevator at school. Also, it’s sometimes hard to keep up with my friends. I do have some dedicated friends that offer to go down the elevator with me and hold books that don’t fit in my backpack.

Recently, my parents applied for a disabled parking sticker so when we go to the store or outdoor music festivals we can park closer to conserve my energy. On days when CMT is hard for me I use CBD medicine. I have it on my nightstand and in my dance bag that way I can apply some where needed and go on with my day.

One thing I found useful is a backpack on wheels so I can roll it in school. My bike is a pink three-wheeler (my friends are super jealous)

Explaining CMT to my friends is hard because when I’m just standing, I seem normal, but I try to explain that this disease makes your hands & feet hurt.

My hope for the CMT Medical Journal project is that once in a million years there is a cure for CMT, so that other people with CMT can have the cure. My case is not as severe. I’m lucky! ”

Thanks Addie you're a SUPERstar and SUPER CMT Model. [Click Here!](#)

If you want to create a visual medical journal that will capture, in sensitive detail, the daily struggles and challenges of living with CMT, please let us know that you are interested by joining the waiting list by emailing registrycoordinator@hnf-cure.org. We’ll be in touch with an invitation to participate when there is an opening.

When a Pandemic Hits... CMT'ers Adapt as Usual!



BY CYNDI MICHENER, LIVING WITH CMT

One of the greatest disappointments for me during the pandemic was the forced cancellation of the 2020 Movement is Medicine™ Summit. The inaugural summit was so motivational and energizing that I was looking forward to the next one before I even returned home from Phoenix. While no virtual event could match the excitement of the in-person event, the March 2021 virtual Movement Is Medicine™ Summit was the next best thing. I am thankful to the HNF and AdventHealth for bringing together so many experts with so much useful information.

Like many people with and without CMT, I have faced challenges with staying active during the pandemic, and mobility challenges add another layer to that difficulty. The information provided during the virtual summit was a great reminder of the particular importance of commitment to exercise, good nutrition, and meditation to those of us with CMT. I hope everyone will have a chance to view the recorded sessions, but I will share a few of the highlights for me.

As a person with severe CMT symptoms, I am always looking for a good, fun workout that I can adapt to my abilities, and the surprise interactive dance routine just after the keynote address met that requirement beautifully. I have to admit that I was glad not to be on camera, but I smiled and sweated the whole way through. The several exercise sessions well spaced through the day were all very well designed for CMT patients and the presenters did a great job showing modifications for people with different abilities. I will definitely be adding some of the moves I learned to my fitness routines.

Meditation is something I have been interested in but hadn't quite known how to get started. I found it very helpful to learn how simple breathing techniques and meditation can help us deal with the levels of stress we all face.

I try very hard to eat a healthy diet, but can never get too many reminders about the importance of eating well. I was not aware that malnutrition and dehydration are common to people with CMT.

Lastly, the slideshows during the breaks and the session on Therapy Across the Lifespan of the Disease reminded me of the many resources that are available to us. [Click Here!](#) Sometimes I get too busy living my life to seek those things out but I want to take advantage of whatever I can find to make life with CMT easier and more enjoyable.

I will be looking forward to the next opportunity to see everyone in person, but again I appreciated this fun and informative event. Stay healthy!

KEEP MOVING & SIGN UP FOR HNF'S MOVEMENT IS MEDICINE™ CLASSES [Click here!](#)



Advent Health

What participants are saying about the Virtual Movement is Medicine™ Summit

This was my first event and it was awesome.

I thought the webinar was fantastic. I am looking forward to joining the exercise classes and using the websites that were given.

Very informative. I found the exercises and nutritional information really helpful e.g. did not realise I should eat protein at every meal.

I am always looking for new exercise ideas, and I definitely learned some moves to add to my workouts. I think the most valuable aspect to me was just the motivation.

I am thankful that you are bringing this to us, most of us really don't know what type of exercise we could do having CMT without getting hurt or doing damage to us.

It was uplifting and motivated me to make changes, one step at a time. Trying the exercises was great.

Coming from the UK — there is not much in the way of support for people with CMT...there is no center of excellence (as far as I know). So it was wonderful to make contact with other CMTers.

As a newly diagnosed person, all of the information I received was invaluable!!! I am so appreciative of all of the information, exercises, and powerful words!!

I think you accomplished your mission 1000 over! Thank you!

5 hours flew by, great organization of content.

The encouragement, enthusiasm and positive attitude were wonderful. I appreciated your ability to stay in your allotted time frames as well.

We were given a wealth of information in an easy to understand manner. The exercises have given me hope for better mobility.

I have CMT and I volunteered for the therapy videos and I didn't realize how many people have cmt or can relate !!! By organizing this summit I have connected with so many people. It motivated me and helped me see past my limitations. I felt empowered and it helped my family and friends see that I do have limits .. it was informative and helpful.

Having experienced the exercises I am now more likely to go online and use recording to possibly even create a program.

A woman is running on a paved path, crossing a finish line marked with blue and red tape. She is smiling broadly with her arms raised in celebration. She is wearing a white athletic shirt with "Team CMT" and a logo, a pink fanny pack, and a blue race bib with the number "A324". She is also wearing black leggings and red running shoes. In the background, there is a large yellow building and a clear blue sky.

"I crossed the finish line at an average pace of 10:40 minutes/mile. Holy Cow, I did it! I did it for me, I did it for my family, and I did it for all who have CMT."

– Team CMT Member:
Gabrielle Putman Blakey

What is Team CMT



Hereditary Neuropathy Foundation

www.movementismedicine.com
www.hnf-cure.org



TEAM CMT was created in 2012 to specifically raise awareness and funds for CMT. We have both abled and disabled members participating in events all over the world to raise funds for CMT research. Our TEAM CMT members proudly wear their Team CMT singlet, t-shirt and/or other swag as they participate in athletic events, work out at the gym and even for a stroll in the park.

Who can join?

Patients, caregivers, friends, and family can join Team CMT.

Is there a fee to become a Team CMT member?

There is no fee to join Team CMT. If members would like to be mailed a Team CMT singlet or t-shirt, HNF requests that a \$25 donation is made to cover the cost.



What type of events can members participate in?

Team CMT members can participate in local bike races, community runs/walks, swim-a-thons, spin-a-thons and golf outings.

How do you fundraise? And will HNF help guide me?

Yes, of course HNF will help guide you. Your success is our success! HNF uses a personalized platform that is user-friendly and will guide you through the process. Donations are made online and donors can also send checks directly to the foundation. The system will send automated thank you letters and you will be notified immediately if anyone donates to your page.

[CLICK HERE TO JOIN TEAM CMT!](#)

Meet...



Traci Williams

Hi, my name is Traci Williams, and I live in Montreal, Canada. I was born with CMT 1A, diagnosed as a child (now in my 50s), and was told I would be permanently in a wheelchair by the age of 20. At 19, I was introduced to yoga and realized I had been meditating naturally on my own about a decade before that. Initially, having a Type A personality, yoga bored me, but my body, even with pain, wanted to do it... and I listened. I have been teaching since 2011 and now have over 500 hours of related training, including in-depth anatomy and physiology. I personally believe strongly in the mind-body-and-spirit connection, so my favorite yoga is Kundalini yoga along with yoga Nidra.

Kundalini yoga is a holistic yoga that incorporates repetitive physical movement and sometimes static postures timed with breathwork, chanting, meditation, and relaxation. It is believed to work directly on the nervous system, glands, and chakra system (the body's energy centers, which largely correspond with the plexuses—bundles of nerves along the spinal cord).

Yoga Nidra is a form of relaxation that brings you to a state between meditation and sleep, tapping into the body's own healing energy.

After doing what most of us know as yoga for years, I took my first Kundalini yoga class in 2001. My initial reaction was, oh, that is so weird, but my body wanted more. Over the years, I have taught many yoga types to many types of students, including teenagers in wheelchairs at a rehabilitation center to women dealing with trauma, depression, and anxiety through a mental health association. Kundalini yoga is not a panacea, but for my own life, it has helped with body and thought awareness, flexibility, strength, balance, perspective, and to celebrate small victories. Given I myself have physical limitations, I have been able to adapt various postures for different levels and different restrictions.



Janice Laurence

Hi, my name is Janice Laurence and I have been a professionally certified teacher in BC, Canada since 1990. I have a MA in education and am passionate about physically integrated dance for all abilities and bodies. I invite you to join me in exploring movement in a joyful and inclusive environment.

Zoom has brought the community of dance into my home and I meet daily with folks of all backgrounds, ages, and genders to learn and move together.

I was born with CMT Type 1A, and have had 14 foot and ankle surgeries. I currently am an ambulatory wheelchair user and have a variety of mobility aids to help me enjoy freedom and independence. As many of you know, living with CMT often means continually finding ways to adapt to a life with unpredictable health challenges and pain. I have been coached by Paralympians and train regularly with Ocean Rehab & Fitness.

I have appeared in several Canadian documentaries, dance films, and live performances as I aim to help dismantle assumptions around living with a disability.

Photo: K. Ho

[Click here to sign up for
Movement is Medicine™ Classes](#)



Paul Fowler

From his middle school teaching days in the 90's to his yearly trips to Thailand to study Thai massage, traditional self-care and meditation to his training at the Functional Medicine Coaching Academy, Paul has been working with people to help them improve their health and well-being for most of his life.

In 2017, when Paul was diagnosed with CMT, he spent a year completely transforming his diet and lifestyle. In that year, much of the nerve pain in his hands and feet receded and though he still has some numbness in his feet, many of the most challenging aspects of the condition have gone away.

After this success, he saw that it was possible to make an impact on CMT through diet and exercise and he wanted to help others who also faced scary and debilitating health challenges, especially people with CMT. That was when he dove into the year-long Functional Medicine Coaching Program. It was the best decision he ever made. He loves working with people using the "coach approach", where they lead and he supports. He has found it endlessly gratifying to see people realize their own power to overcome things that they never before thought possible.



Laura Mayer

Hi my name is Laura Mayer. I am very excited to be here now. I bridge my clinical expertise as an Occupational Therapist, deep intuitive knowing and body of proof that healing and wellness happens when we pay attention and love ourselves no matter what! I am also an author, teacher and kickass attitude adjuster.

My greatest gifts did not come from schooling but rather personal life experience. Diagnosed at fifteen in NYC with a progressive, degenerative neurological disease (Anterior Horn Cell Disease) I was told I would lose a millimeter of muscle strength a month --starting in my hands and then progress throughout my body until my death at 40. At 65 I am thrilled to be of service!

My hands became the focal point of my life. I had to learn how to compensate, monitor energy expenditure and do the best I could. I know all about hands -- and I am here to assist you to know and love your hands.

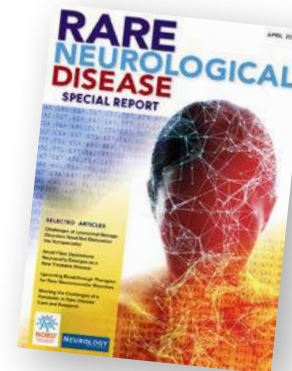
As a practicing Occupational Therapist for over three decades, and now as a Healing Activator, my passion and purpose is to help others to activate their own healing within, helping YOU master YOUR life as stress-free as possible! Through **Movement is Medicine™** we will put some *fun* into hand *functionAbility*.

Give Yourself a Hand-From One Loving Hand to the Other will help you navigate how to maintain flexAbility, minimize energy expenditure, do your own purposeful activity analysis for maximal function and most importantly, open up your heart to loving you as you are.

I am wide open to your questions, needs and concerns. My wish is that these classes will be interactive to give you the most for your invested time and energy.

Website: laurajmayer.com **Facebook:** [laura.mayer.52035](https://www.facebook.com/laura.mayer.52035)
Instagram: [paint_intuit](https://www.instagram.com/paint_intuit)

[Click here to sign up for
Movement is Medicine™ Classes](#)



HNF Continues Raising Awareness of CMT among Neurologists and Health Care Providers

For the seventh year, we are proud to be part of the annual **Rare Neurological Disease Special Report**—a widely-read supplement mailed out with the April, 2021 issue of *Neurology Reviews* and distributed at all major neurology conferences throughout the year.

We are diligently educating neurologists and health care providers about CMT on your behalf.

Neurology Reviews is the first and original news source in neurology and has a history of providing independent, unbiased news to neurologists and clinicians interested in neurosciences.

This year's Special Report features the Pharnext PREMIER Trial of PXT3003 CMT1A Clinical Trial, plus so much more...

Featured articles about CMT and HNF's related initiatives include:

- Linking neurologists to clinical trial opportunities ([pg. 8](#))
- Pharnext PREMIER Trial of PXT3003 CMT1A Clinical Trial - Now Screening for Participants ([pg. 9](#))
- The CMT Care Pathway for Doctors and Patients ([pg. 61](#))

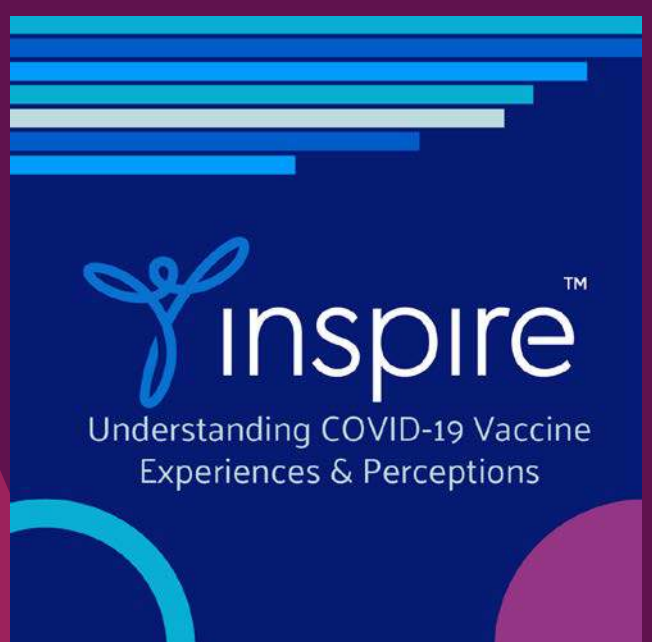
Be sure to access the full Rare Neurological Disease Special Report download [here](#). If you would like your own printed copy, please contact Elizabeth Katz, the publisher of NR, at ekatz@mdedge.com or call 973-224-7951.

As with all of HNF's programs, we rely on donations and support from our industry sponsors, government grants, and **YOU** that want to help us with our medical outreach initiatives.

COVID-19 Vaccine Survey

Our partner, Inspire™ is conducting an ongoing survey about the COVID-19 vaccine. We invite you to contribute to a broader understanding of vaccine perceptions, symptoms, and access in the CMT community by **taking the survey**.

[CLICK HERE!](#)





HNF Continues Partnership with AdventHealth: CMT Uplift – Monthly Support Group for Patients & Caregivers



Meet Laura Russell, MSW, LCSW Neuromuscular Medicine, Social Worker, AdventHealth. Laura is a licensed clinical social worker who has demonstrated experience providing counseling and various intervention techniques to children, adults and geriatric clients with a proven ability to deliver quality service under high pressure and resource constraints, while continuing to uphold core values and being sensitive to multi-cultural, generational, and diverse populations.

Join Laura's monthly virtual peer-to-peer interactive support groups for CMT via Zoom. These support groups will focus on personal experiences and living with CMT.

Charcot-Marie-Tooth (CMT) Support Group

First Tuesday of every month from 1-2pm EDT.

This is a virtual space where you can discuss the emotional and physical challenges of living with CMT. No matter what age, stage or relationship to CMT, there is always comfort in staying informed and having a safe environment to express challenges and learn coping techniques from peers. Our Support Group addresses many topics and symptoms of the disease as well as coping with changes of chronic illness. In this group, you will hear from others at varying ages, stages and relations to the disease as well as from experts in the field including Neuromuscular Medicine Specialists.

REGISTER: [Click here!](#)

In Sickness & Health Care Partner Support Group

First Thursday of every month from 12:30pm-1:30pm EDT.

Has your loved one been diagnosed with a neuromuscular disorder? You may have questions and/or just want to talk to others who are also on the same journey of helping a loved one navigate life with a neuromuscular condition - while managing your own life, needs and stress. Join us for an hour of honest conversation, possible tears, and lots of laughs. This is a casual, come as you are, welcoming setting!

REGISTER: [Click here!](#)

LEARN MORE ABOUT ADVENTHEALTH NEUROSCIENCE INSTITUTE: [Click here!](#)

Top 7 Reasons Why Patient Organizations are the Secret Weapon to Future Treatments



CONTRIBUTED BY: ESTELA LUGO, HNF, MANAGER OF PROGRAMS

I wish I could say there was a time in my life when I felt “normal,” but there really isn’t. Some of my first memories include sitting on the number rug in kindergarten and showing off my new plastic leg braces to friends.

It was 1984 when doctors at Nassau County Medical Center in New York broke the solemn news to my parents. Their 4-year-old daughter was not just a clumsy kid; she had a neuromuscular disease called Charcot-Marie-Tooth (CMT). The only information neurologists could give them was that my hands, legs, and feet would progressively weaken over time and that there were no treatments or cure. Two years later, my little sister began showing symptoms and was also diagnosed with the same fate. You can imagine their shock to learn of our prognosis. There was no history of CMT in our family, and my parents were as strong and healthy as they come. Where many families could have fallen victim to shame, self-pity, and despair, my incredible parents decided to become overnight advocates.

From that point forward, our family became heavily involved in raising awareness and research funding as the official Muscular Dystrophy Association (MDA) poster children for Long Island, N.Y. We would bounce around from one event to another, meeting kind and compassionate individuals along the way whose missions aligned with our own; to cure debilitating diseases like CMT. These were some of the most significant years of our lives. One of the most poignant parts of it all was the healing it provided for our family. Advocacy quite literally turned our pain into purpose

while also building up our confidence. We grew up experiencing the importance of volunteer work, service, and community. And we developed a deep sense of humility and gratitude for the challenges we did have because we now personally knew many families whose daily battles dwarfed our own.

Fast forward to 2021, and advocacy is now my full-time career. After volunteering for the Hereditary Neuropathy Foundation for eight years, I joined their dedicated staff as program development manager in 2018. Being part of this incredible organization has entirely turned my prior notions of patient advocacy on their head. I’ve witnessed the impact a few dedicated individuals can have on an entire disease community and the drug development process. I’ve seen mothers become empowered disease experts to save their children’s lives, funding and championing breakthrough gene therapy treatments. Long gone are the days where patients sit back and wait for a cure.

In contrast to my younger years, Patient Advocacy Organizations are anything but “poster children.” Instead, we are now the driving force behind future treatments and therapies.

Here are seven reasons why:

1. We gather vital patient data through patient registries, surveys, and studies.
2. We source and implement emerging technologies to accelerate clinical research and trials with AI, apps, and innovative digital technologies.
3. We consult with and educate the FDA through Externally-Led Patient-Focused Drug Development Meetings and Critical Path Innovation Meetings.
4. We are the experts of the disease experience and its impact on daily life. We understand and evaluate benefit/risk as well as critical endpoints for clinical trial success.
5. We have our finger on the pulse of the patient community with networks covering the globe, making CMT and other rare diseases not so rare. Our vast infrastructure and programs allow for fast and efficient recruitment for clinical trials.
6. We have earned our community’s trust through extensive support and transparency as patients, families, and caregivers ourselves.
7. Our passion, drive, and sense of urgency are unmatched as we are personally affected by our disease and understand the true cost of each day without treatments.

Get Involved!

NEUROTOXIC DRUG CAUTION

Fluoroquinolones (FLOOR·o·QUIN·o·loans)

- Levofloxacin (Levaquin)
- Ciprofloxacin (Cipro)
- Moxifloxacin (Avelox)



Want an easy way to spot a fluoroquinolone? — The generic name will end in “**floxacin**”

Fluoroquinolones are a class of antibiotics used to treat a wide variety of infections including urinary tract infections, pneumonia, skin/bone infections, sexually transmitted infections etc. These antibiotics kill bacteria by inhibiting DNA synthesis within the bacteria cell. While effective at treating these ailments, they are also associated with a significant risk of sensory and motor peripheral neuropathy including axonal neuropathy and Guillain-Barre syndrome (a rare disorder in which your body's immune system attacks your nerves). Several case reports have indicated a very slow recovery and/or a permanent state of disability/neuropathy. How these antibiotics damage the peripheral nervous system isn't completely understood.

This risk exists for anyone receiving a fluoroquinolone but is **especially increased in patients with CMT**. Other factors that have been shown to increase your risk include being over 60, being male, and longer durations of therapy. In fact, a recent study from 2019 found the relative incidence of peripheral neuropathy increased by approximately 3% with each additional day of oral fluorquinolone use. However, other case reports demonstrated neuropathy presenting as early as the first day of therapy. Additionally, neuropathy symptoms can present days to weeks after the completion of therapy.

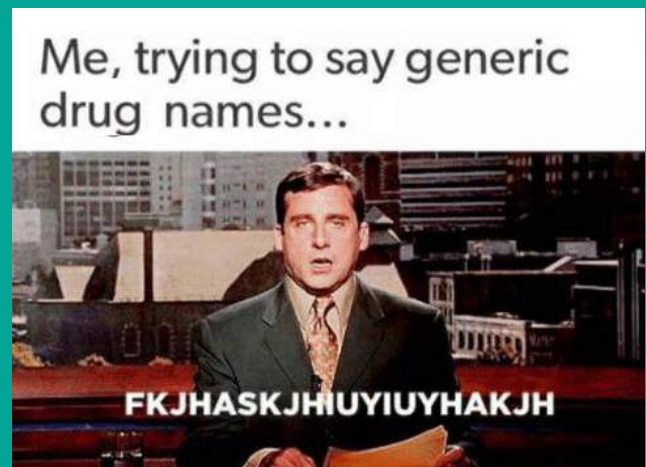
Therefore, whenever possible, these antibiotics should be avoided in CMT patients. Luckily, other antibiotics are available and are often just as effective (see below)

CONTRIBUTOR: Corinne Weinstein, Clinical Oncology Pharmacist at Cancer Center of Colorado-Good Samaritan, Denver, Colorado

Antibiotics with minimal to no risk of neurotoxicity (not all inclusive)

- Amoxicillin
- Amoxicillin/Clavulanate (Augmentin)
- Ceftriaxone
- Cefuroxime (Ceftin)
- Cefpodixime
- Cefdinir
- Cephalexin (Kelfex)
- Doxycycline
- Trimethoprim/SMX (Bactrim)
- Azithromycin
- Clindamycin

****Of note, only systemic fluoroquinolones (i.e pills, IV) have been associated with an increased risk of peripheral neuropathy. Ear drops, eye drops, or lotions that contain a fluoroquinolones are not likely to increase this risk****





“Grow or Mow” A Sheer Success

The Covid flow has been strong for many, especially for Brandon Daily, who hasn't cut his hair since pre-pandemic! Brandon couldn't decide to let it grow through the rest of 2021 or shave it off. That's when his wife, Shelley, thought of a fun way they could let others decide while raising research dollars for their niece, Alana, who lives with CMT GDAP1 (Type 4a). **They would host a virtual 'Grow or Mow' fundraiser** with each \$1 vote going towards CMT research.

One week later, the RESULTS WERE IN! 1173 GROW votes to a staggering 1690 for MOW. It was a chilly morning to get a buzz cut, but Brandon was a trooper as Alana wielded the clippers for the first time.

*"We're so thankful to everyone who voted! Our fundraiser total, including donations and matched money, was **\$6241!** HNF made the entire process turnkey and effortless with a customized webpage, promo images, and voting updates...the hardest part was sweeping up all of Brandon's hair!"*

– Shelley

Luxurious Locks Wanted!

Nominate someone for the next CMT “Grow or Mow” fundraiser and choose where your research funds go! Email us at info@hnf-cure.org and ask how to get started!



Natalia Salejko New Board member

Natalia Salejko is a happy wife and mom of two wonderful girls, one of whom has CMT4A. Her professional background includes; marketing, medical environments, and managing operations for an international company headquartered in London. Currently, Natalia is the office manager for the largest e-commerce company in Poland, Allegro. After living in the UK for 14 years, her family moved to Poland. Medicine has always been one of her greatest passions. Her family and growing team are laser-focused on finding a cure through aggressive fundraising through many channels.



Faces of CMT Rare Disease Day 2021 Photography Competition

To mark Rare Disease Day 2021, the inaugural “Faces of CMT Photography Competition” was supported by Pharnext, to raise awareness of the challenges of living with CMT and the strategies used by individuals to overcome them.

The competition was open to everyone around the world, for those with CMT or those living alongside someone with CMT. The level of response from all areas of the world was inspiring and the submissions and stories were amazing.. To celebrate everyone’s participation in the competition a “Faces of CMT Winner & Finalist” digital book was published of all of the creative, impactful and heartfelt submissions.

You can view the photo book on our website here: [Click!](#)

Ultimate CMT Amazon Wish List

HNF’s focus is to support people living with CMT and their families with critical information to improve quality of life and we are dedicated to finding treatments for CMT. We provide a strong, organizational voice to those living with CMT all over the world.



The HNF team and supporters came up with a list on Amazon that CMT’ers currently use to make their life with CMT easier!

We hope you find this list helpful and choose HNF as your AmazonSmile Charity moving forward.

View HNF’s Ultimate CMT Amazon Wish List [here!](#)

Simply start your shopping at smile.amazon.com/ch/13-4137654 to confirm "Hereditary Neuropathy Foundation Inc" as your charity of choice, and AmazonSmile will donate a portion of your eligible purchase price to our organization.



New Center of Excellence: University of Washington Medical Center, Seattle, Washington

DR. NASSIM RAD



Q: TELL US ABOUT YOURSELF:

I am a board-certified rehabilitation medicine physician (completed my residency at Northwestern University) and a board-certified neuromuscular medicine physician (completed my neurology neuromuscular fellowship at the University of Michigan). I am also certified in electrodiagnostic medicine.

Q: WHY IS CMT YOUR PASSION?

I truly enjoy educating patients and helping them become experts in their medical care and CMT is a disease that requires just that! I work with adult patients from diagnosis and throughout their life to manage symptoms so that they can live their most functional life. Education in CMT is key as the complex presentations and genetic work-up can be overwhelming to patients. To add to this, the progressive nature of CMT can be frustrating and disappointing. I enjoy showing patients how they can overcome these barriers to increase mobility, reduce pain and live a fulfilling life.

Q: TELL US WHAT PATIENTS WILL EXPERIENCE WHEN THEY COME TO YOUR CENTER?

Patients are typically seen on Wednesdays. They will meet with a physician (either rehab or neurology) that has expertise in CMT. They have access to our respiratory therapists and liaisons as well as our nurse coordinator for any issues that come up. Same day PT and OT evaluations can also be arranged. We work closely with our genetic counselors and help plug patients into any necessary referral services including a visit with our orthotics and prosthetics department. Given the current state of the pandemic, telemedicine visits

are available, and the multidisciplinary aspect of the clinic is limited to avoid unnecessary exposure.

Q: WHAT DO YOU LOVE MOST ABOUT YOUR PRACTICE?

My patients; I have the opportunity to work with some of the most resilient people! Additionally, my colleagues and staff are a pleasure to work with. They are experts in the field and put patients first.

Q: HOW CAN PATIENTS MAKE AN APPOINTMENT AT YOUR CENTER?

CMT Scheduler Phone Number:
206.598.0058

ASK THE EXPERT

Dear Lainie,

One of my biggest frustrations with CMT is that my fingers don't do what they should and what I need them to do on the go. It's embarrassing when I can't pick up a quarter off of a counter at the grocery store or when any small object drops on the ground. What do you do? Are your hands bad? HELP!

From Lisa Peterson

Hi Lisa:

I get it and completely understand your frustration! You do not realize how many everyday tasks require the use of a pincher grip until you don't have one. Am I right? As you probably know, when you have Charcot Marie Tooth Disease and fine muscle weakness in your hands due to neuropathy, it can be very difficult to pick up dropped change, crumbs of food, and basically any other small and flat item.

Although I try to be positive and live my best life despite living with CMT, it's the small, hand-related tasks that can be the most frustrating and get me off my groove. Although I do own one of those long grabber sticks that help people with a weak grasp reach and pick up things, my problem is that I just have a weak pincher grip and no difficulty bending down. Frankly, I can use all the exercise I can get! 😂

To solve the challenge of picking up small, flat objects, I discovered that I could repurpose a lint brush to be a mini grabber device. They come in a ton of sizes and the handles are typically large and easy to hold onto. As an added bonus, they are super cheap! You can find them at any dollar store, or here are the mini ones I buy on Amazon and keep in my purse. I hope this little hack helps! Let me know what you think.



Xoxo
Lainie

Lainie Ishbia, MSW
Disability Speaker & Blogger
Trend-ABLE



Do you ever wish you could have direct access to a neurologist for your CMT questions? Now you can! HNF continues to feature real questions from CMT patients across many topics. Submit your questions from our site to the Directors of the HNF Centers of Excellence, Dr. Jafar Kafaie for St. Louis University School of Medicine, St. Louis, MO, and Dr. Florian Thomas for Hackensack University Medical Center & Hackensack Meridian School of Medicine, Hackensack, NJ

Q: My calf muscles have wasted away, affecting my balance and walking. Can these muscles ever be restored?

A: Dr. Florian Thomas:

Thank you for sharing that question. In CMT the calf muscles waste away because the nerve fibers to the muscle are damaged. In order to avoid this the disease process in the nerve fibers must be halted or reversed.

That is the goal of numerous CMT drug trials in various stages across the globe. Some of these experimental drugs target only specific genes that are mutated in CMT, while others may benefit patients with different CMT subtypes. While we have had encouraging news from some of them, none are yet close to FDA approval

HAVE A QUESTION?

www.hnf-cure.org/ask-the-expert



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 patients and families with inherited neuropathies.

ARIZONA

Barrow Neurological Institute
350 West Thomas Road
Phoenix, Arizona 85013
Contact: Rose Anaya
602-406-2704

ARKANSAS

Arkansas Children's*
Little Rock, AR 72202-3591
Contact: Dr. Aravindhan
Veerapandian
Primary Care Appointments:
501-213-1883
Specialty Care Appointments:
501-819-3520
Hospital *Pediatric Center
of Excellence

CALIFORNIA

Cedars-Sinai Medical Center
Los Angeles, CA
Contact: Tara Jones
tara.jones@cshs.org
CMT Clinic line 310-423-4268

Stanford Neuroscience Health Center
Neuromuscular Clinic
Palo Alto, CA
Contact: Jennifer Fisher
jnfisher@stanford.edu

UCLA Department of Neurology
300 Medical Plaza, Suite B200
Los Angeles, CA 90095
Contact: Dr. Halabi Anasheh Office
Telephone: 310-794-1195
(note: request to be scheduled with
Dr. Halabi)

COLORADO

Children's Hospital Colorado*
Contact: Alison Ballard at 720-777-
8723 or call the clinic schedulers at
720-777-2806

CONNECTICUT

Hospital for Special Care
New Britain, CT
Contact: Angel Preece, RN
860-612-6305 APreece@hpsc.org

FLORIDA

AdventHealth Neurology at Winter Park
1573 W Fairbanks, Ste 210
Winter Park, FL 32789
Contact: Nivedita Jerath MD, MS
407-303-6729
Orl.neuromuscular.medicine@
adventhealth.com

University of Florida Health
3009 Williston Road
Gainesville, FL 32608
Contact: Melissa Hines
352-294-5000 mhines@ufl.edu

University of Miami
Professional Arts Center (PAC)
1150 NW 14th Street, 6th Floor
Miami, FL 33136
Contact: Roberto Fernandez
r.fernandez5@med.miami.edu

ILLINOIS

Ann and Robert H. Lurie Children's Hospital of Chicago*
225 East Chicago Avenue
Chicago, IL 60611
Contact: 312-227-4471

KANSAS

University of Kansas Medical Center
Kansas City, KS
Contact: Andrew Heim 913-945-9926
aheim2@kumc.edu

MASSACHUSETTS

Brigham and Women's Hospital
Boston, MA
Contact: Kristen Roe 617-525-6763
kroe@partners.org

MICHIGAN

University Of Michigan
Ann Arbor MI
Contact: Keianna Banbury
734-763-2554 kbanbury@med.umich.edu

MINNESOTA

University of Minnesota Health
Maple Grove, MN
For Research Studies: 612-624-7745
CNRU@umn.edu
For Clinic Appointments:
763-898-1080

MISSOURI

St. Louis University Medical Center
St. Louis, MO
Contact: Mrs. Molly Labrier
314-977-6177
molly.labrier@health.slu.edu

MU Health Care
Columbia, MO

Contact: Dr. Raghav Govindarajan
573-882-1515 govindarajanr@health.
missouri.edu

NEW JERSEY

Hackensack University Medical Center
Hackensack, NJ
Contact: Florian Thomas, MD, PhD
551-996-8100
Annerys.Santos@HMHN.org

Atlantic Health System*

Morristown, NJ
Contact: Dr. Jahannaz Dastgir
973-971-5700
jahannaz.dastgir@atlantichealth.org

NEW YORK

Columbia University
New York, NY
Contact: For clinical appointments
Allan Paras 212-305-0405
For research studies 212-305-6035
ap3476@cumc.columbia.edu

NORTH CAROLINA

Dr. Rebecca Traub
University of North Carolina
194 Finley Golf Course Road, Suite 200
Chapel Hill, NC 27517
Contact: For clinical appointments
984-974-4401
Referral Fax: 984-974-2285

Atrium Health Neurosciences Institute-Charlotte

1010 Edgehill Road North
Charlotte, NC 28207
Contact: The phone number for
appointments is 704-446-1900. Ashley
Clyburn is our incoming referrals
coordinator and referrals can be faxed
to 704-355-5650

TEXAS

Austin Neuromuscular Center
3901 Medical Parkway, Ste. 300
Austin, TX 78756
Contact: Yessar Hussain, MD
512-920-0140
Website: [austinneuromuscle.com/
contact](http://austinneuromuscle.com/contact)

WASHINGTON

St. Luke's Rehabilitation Institute
Spokane, WA
Contact: Ann Cooper 509-939-8079
coopera@st-lukes.org

University of Washington Medical Center

1959 NE Pacific Street
Seattle, WA 98195
Contact: Dr. Nassim Rad
CMT Scheduler (206)598-0058

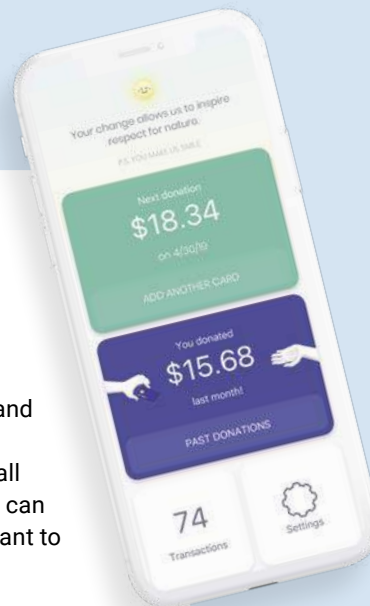
Help Support HNF with the RoundUp app!

HNF has partnered with an app called RoundUp that allows you to round up and donate the change from your credit or debit card purchases to support us – all automatically and without hassle. You can even cap the maximum amount you want to donate in a given month. [CLICK HERE](#).

If you would be willing to support us in this way, simply download the app or use the web version at roundupapp.com. You will be able to create an account and choose us when prompted to select the organization you will support. Also, we would love for you to spread the word to other individuals who may be interested!

Please let us know if I can answer any questions
courtney@hnf-cure.org

Thanks,
Courtney



HEREDITARY
NEUROPATHY
FOUNDATION

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New York, NY 10128
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HNF
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Events

Get Involved and Join us at an Upcoming HNF Event

Chicago Golf Event

September 27, 2021

Jaxson Crusaders Clay Shoot

October 16, 2021

TCS New York City Marathon

November 7, 2021



**PHAR
NEXT**

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