



HEREDITARY  
NEUROPATHY  
FOUNDATION

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# cmtupdate

## September Awareness Month

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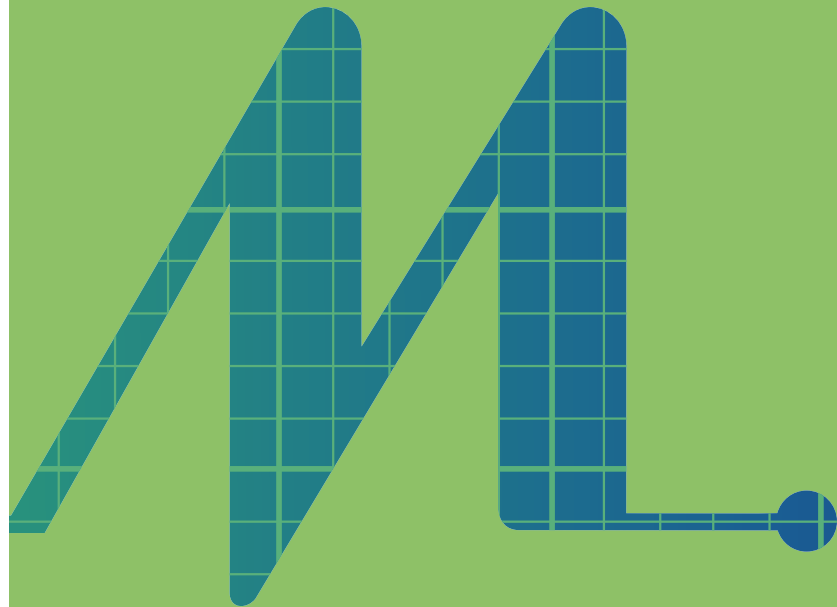
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#### **DESIGN**

BolleDesign.com

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](http://www.hnf-cure.org)



# CMT UPDATE SUMMER 2019



**Allison T. Moore**

Founder/CEO, Hereditary Neuropathy Foundation

Dear Friends,


As the summer is wrapping up, HNF is preparing for the second half of 2019. We've launched several new programs that are continuing to provide support to our CMT patient and family community, as well as three new research initiatives that show promise for rarer types of CMT. Our new ***Movement is Medicine™*** program is breaking new ground in identifying specific exercises to accommodate ALL patients with CMT, regardless of the level of their symptoms. This year's annual Summit is being held at Ability360 in Phoenix, AZ, on November 8 and 9. Registration is filling up quickly, so if you want to join us, don't wait!

**VISIT: [www.movementismedicine.com](http://www.movementismedicine.com)**

With **September CMT Awareness Month** in full swing, we are excited to bring back our Spin Cycling events to spread awareness and raise funds to support our research initiatives. We're calling on our dedicated HNF members and friends to support and spin with us throughout the month of September. There are so many ways to get involved.

Finally, I am eternally grateful to our donors, industry partners, and private foundations for their continued support of our patient programs, the **Global Registry for Inherited Neuropathies (GRIN)** and our innovative **Therapeutic Research In Accelerated Discovery (TRIAD)** program.

***Together, we are stronger!***

A handwritten signature in cursive script that reads "Allison T. Moore".

# SEPTEMBER AWARENESS MONTH SPIN WITH US!

Get inspired and connect with patients, friends and loved ones who are all on the same journey, to cure CMT! HNF is spinning into September Charcot-Marie-Tooth (CMT) Awareness Month 2019 with a year-round, nationwide awareness and research initiative to support our Therapeutic Research In Accelerated Discovery (TRIAD) Program.

It's a unique opportunity for our community to come together and celebrate, and raise awareness and funds for CMT research. Whether you have spun with us before or are a first-time participant, our spins will empower you and leave you energized and optimistic about the future. To date, HNF has funded over \$2 million in research. In order to continue to support our patient community and expand our CMT research initiatives, we need your help!

We are bringing back our wildly successful spin events even bigger and better as we launch Team CMT Spin from coast to coast. Registration for Santa Monica, CA (9/21) & Boston, MA (9/28) is now open!

**VISIT:** [www.hnf-cure.org/cmt-events/spin-a-thon/](http://www.hnf-cure.org/cmt-events/spin-a-thon/)

**Want to be part of the action and have fun too?**  
**We need hosts for more Spin events across the country!**

## Here's how!

1. **Host** - Raise awareness, make new friends and create an impact for everyone with CMT. The HNF is looking for dedicated volunteers to host Team CMT Spin events nationwide.
2. **Participate** - Register for a Team CMT Spin in your area or volunteer to work at the event!
3. **Donate** - Select a city to support! Any size donation takes us one step further in Charcot-Marie-Tooth research.





Bringing Back

# TEAM CMT SPIN

## HERE ARE SOME OTHER WAYS YOU CAN GET INVOLVED!

### HNF's New Auto Donation Program

What seems like an insignificant amount adds up to real CHANGE when we work together with a common mission. Instead of spending \$10 on coffee one week, set-up a charitable donation to HNF.

Sign up for HNF's new auto donation program and we'll send you a set of Bombas Socks to keep your feet as warm as your heart!

**VISIT:** [www.hnf-cure.org/coffee-campaign](http://www.hnf-cure.org/coffee-campaign)

### Tattoo Your Heart Out

Make a \$10 donation to HNF and receive a 3-pack of tattoos to spread awareness for CMT. Don't forget to tag us on social media.

**VISIT:** [weblink.donorperfect.com/tattoocampaign](http://weblink.donorperfect.com/tattoocampaign)

#HNF4CMT #CMTWeGotThis

### September Awareness CMT Kits

HNF has designed a September Awareness Month kit to assist you with spreading the word about CMT. Learn, laugh, and spread awareness with each kit.

**VISIT:** [www.hnf-cure.org/cmt-awareness-month](http://www.hnf-cure.org/cmt-awareness-month)

### Docs That Rock

Have excellent doctors or healthcare providers? Nominate your Docs That Rock and tell us how they meet your needs as a CMT patient. We'll share your submission on our dedicated web page for your fellow patients nationwide!

**VISIT:** [www.hnf-cure.org/docs-that-rock](http://www.hnf-cure.org/docs-that-rock)



### Shop Our Store

Spread CMT awareness with style! We've designed new T-shirts, phone cases, and more. 100% of the profits will go to HNF's TRIAD program for CMT research.

**VISIT:** [www.cafepress.com/cmtwegotthis](http://www.cafepress.com/cmtwegotthis)

### Follow Us On Social Media

Like, comment, and share the latest HNF CMT news and events on all your social media channels with the hashtag #CMTWeGotThis. Don't forget to print your Selfie Card and post pictures, making sure you tag a friend.

**VISIT:** [www.hnf-cure.org/social-media-outlets](http://www.hnf-cure.org/social-media-outlets)

### Reach Out To Your Representatives

Be the change you wish to see. Find your local representative through our linked directory and then download, print and mail our custom CMT advocacy letter to them.

**VISIT:** [www.hnf-cure.org/advocacy](http://www.hnf-cure.org/advocacy)

### Band Together For CMT

Receive your exclusive set of resistance bands with our guide book of CMT-specific exercises with every \$25 donation.

**VISIT:** [www.hnf-cure.org/resistance-bands-cmt](http://www.hnf-cure.org/resistance-bands-cmt)

### More Ways To Get Involved

HNF has several ways you can make a contribution to help raise awareness and funds for CMT research.

**VISIT:** [www.hnf-cure.org/get-involved-for-charcot-marie-tooth-awareness-month](http://www.hnf-cure.org/get-involved-for-charcot-marie-tooth-awareness-month)

# HNF is Now Recruiting Patients for New Gene Therapy Research Projects

Not all types of CMT can be cured with Gene Therapy, but there are many types that can. HNF is leading efforts in identifying those types and spearheading initiatives with a collaborative team of experts in the field of gene therapy.

HNF has selected its first three types of CMT based on their potential for success...and we need your help! If you are a patient or know someone with the following CMT types, contact [allison@hnf-cure.org](mailto:allison@hnf-cure.org).

## CNTnap1

This rare and fatal type of CMT is recessive and leads to a loss of the CNTNap1 gene and its protein product, CASPR which disrupts the myelinating cells from the signal transmitting nerve cell axons. This has profound negative effects on the development and maintenance of many nerve fibers in the central and peripheral nervous system.

James was born with CNTnap1 and unable to breathe. He was immediately intubated. After he was stable, the NICU team at the local hospital decided it was best to send James that night to a Children's Hospital specializing in rare diseases.

The clock is ticking as HNF works hard to raise funds to save this little boys life and others like him.

### TO WATCH MORE OF JAMES' STORY:

[www.jamescure.org](http://www.jamescure.org)



The McCarthy Family

## CMT4 (GDAP1)

CMT4 is a subtype of CMT that is inherited in an autosomal recessive pattern.

Estela Lugo, HNF Medical Outreach Director and her sister, Melissa were diagnosed with CMT at age 3. For many, many years they did not know their type. Previous genetic tests were inconclusive up until recently.

Working for HNF, shed light on the importance of getting a genetic



diagnosis and that new genes are being discovered all the time. Last year, Estela and Melissa finally received a confirmed diagnosis for CMT4 GDAP1. It was only natural that Estela join in the effort of spreading the word and participating with a heartfelt testimony at the Externally-led Patient Focused Drug Development Meeting for the FDA last year.

### TO WATCH ESTELA'S TESTIMONY:

[hnf-cure.org/cmt4-gdap-gene-therapy](http://hnf-cure.org/cmt4-gdap-gene-therapy)

## CMT6 (c12orf56) “One gene, two diseases”

What does this mean? C12orf56 can present as two different types of diseases called CMT6 and Leigh's Syndrome. Both diseases are caused by dysfunction of mitochondria, the energy factory of our cells, and are progressive and, many times, devastating.

CMT6 is a complex type of CMT because it is associated with many other features in addition to the typical CMT symptoms such as delayed learning, blindness, language impairment and shorter life expectancy. Some patients with mutations in the C12orf56 associated with severe mitochondrial dysfunction can also present an aggressive central nervous system disease called Leigh's syndrome. Symptoms are frequently triggered by a viral infection and include developmental delay or regression, loss of coordination, vision and hearing loss, memory disturbance and low or weak muscle tone.

HNF has been working hard to raise funds for Zachary Houliars, a young man, age 21 and others like him to find a

treatment. Despite Zachary challenges and being legally blind, he has excelled in his community as a sport star coaching for his high school team and is continuing his passion at the college level. This past winter, Zach caught the flu and experienced a severe decline of his CMT which led to the diagnosis of Leigh's Syndrome. It's only now that we have learned, that CMT6 and Leigh's Syndrome share the same gene, but Leigh's syndrome presents more severely.

How could this be? Zach was hospitalized and almost died from severe mitochondrial dysfunction. Unable to swallow, speak and even move. He spent months in the hospital and is slowly recovering back at school.

HNF, in collaboration with Robert Burgess, PhD at Jackson Labs, Steven Gray, PhD at UT Southwestern and Mario Saporta, MD, PhD, MBA at the University of Miami, are working diligently on developing a gene therapy for these diseases.

**LEARN MORE:** [www.curecmt.org](http://www.curecmt.org)

**“As genetic therapies quickly become a real option for CMT patients, these new HNF projects are very exciting for patients, physicians, and researchers alike”**

– **Dr. Stephan Züchner, MD, PhD**  
Professor for Human Genetics and Neurology Chair, Dr. John T. Macdonald Foundation Department of Human Genetics Co-Director, John P. Hussman Institute for Human Genomics University of Miami Miller School of Medicine and Founder, The Genesis Project, a key collaborator of HNF's Gene Therapy Research initiative



## *Movement is Medicine™*

Movement is Medicine program, an innovative research initiative to bring exercise modalities to all patients with CMT. These studies have the potential to be an adjunct therapy to improve functional outcomes and endpoints in future CMT patient clinical trials.

**REGISTER NOW! [WWW.MOVEMENTISMEDICINE.COM](http://WWW.MOVEMENTISMEDICINE.COM)**





## Orphan Disease Center **Million Dollar Bike Ride** Pilot Grant Program

The ODC MDBR Pilot Grant Program provides a one-year grant to support research related to a rare disease represented in the 2019 Million Dollar Bike Ride. HNF reached its \$30,165 goal which was generously matched by the Orphan Disease Center. We want to thank all our riders and donors for participating. The grant will be a 1 year award with an optional 6 month no cost extension for a one time award of \$60,330 starting January 1, 2020.



**APPLY  
TODAY**

[Click Here](#)

### Eligibility

All individuals holding a faculty-level appointment at an academic institution or a senior scientific position at a non-profit institution or foundation are eligible to respond to this RFA.

### Letter of Interest Instructions

Please visit the ODC website ([orphan-disease-center.med.upenn.edu/grant-opportunities](http://orphan-disease-center.med.upenn.edu/grant-opportunities)) to submit your Letter of Interest (LOI), which can also be found here: [orphan-disease-center.submittable.com/submit](http://orphan-disease-center.submittable.com/submit)

**This one-page LOI is due no later than Monday, September 23, 2019 by 8pm (EST).**

**Click RFA guidelines and scroll down to #7 for details for CMT grant.**

### Full Application Instructions and Review Procedure

NOTE: Full Application is by invitation only after review of Pre-Application

### Proposal Due Date

Thursday, October 24, 2019 no later than 8pm (EST)  
Full application documents are to be uploaded on our website, by invitation only.

Gretchen Cappiello, Allison Moore  
and Matt Downing



# HNF Presents Research Findings at Three Prestigious Conferences.



## Charcot-Marie-Tooth 1A (CMT1A) and Impaired Patient Mobility-Expressions, Remedies and Impact on Quality of Life.

**ISPOR NEW ORLEANS MAY 2019  
PERIPHERAL NERVE SOCIETY  
MEETING JUNE 2019**

**Conclusion:** Patient mobility was the #1 issue for patients with CMT1A; it has significant impact on the multiple aspects of a patient's QoL.

Patient phenotype, like curled toes, pes cavus and muscle atrophy contributes directly to impaired mobility. Patients that do not opt for corrective surgery use multiple modalities to assist with mobility, with bracing and orthotics used by large cohort of patients.

Pain and fatigue are also a serious by-product of patient mobility issues, along with frequent falls.

**[CLICK HERE TO VIEW POSTER](#)**

[www.hnf-cure.org/wp-content/uploads/2019/08/IPSOR-Poster-FPT051619-1-1-1.pdf](http://www.hnf-cure.org/wp-content/uploads/2019/08/IPSOR-Poster-FPT051619-1-1-1.pdf)

## The Path to Diagnosing Charcot Marie Tooth Disease: The Patient Experience

**ISPOR NEW ORLEANS MAY 2019  
PERIPHERAL NERVE SOCIETY  
MEETING JUNE 2019**

**Conclusion:** Although many CMT patients obtain a confirmed diagnosis of their disease within 12 months of manifesting symptoms, a significant cohort of patients can take several years to obtain an accurate diagnosis. This is particularly prevalent in older patients that manifest symptoms later in life.

Given the significant patient population under 20 years of age that are diagnosed, pediatricians were underrepresented in our patient survey as the HCP that first recognized patient symptoms.

Although the majority of patients received their diagnosis via genetic testing, cost of testing was noted as a barrier.

**[CLICK HERE TO VIEW POSTER](#)**

[www.hnf-cure.org/wp-content/uploads/2019/08/Diagnosis-PNS-poster-.pdf](http://www.hnf-cure.org/wp-content/uploads/2019/08/Diagnosis-PNS-poster-.pdf)

## Comparison of the Risks and Benefits of Medical Cannabis in Charcot-Marie-Tooth (CMT) and Hereditary Neuropathy Pressure Palsies (HNPP) versus Chronic Pain Patients

**EASTERN PSYCHOLOGICAL  
CONFERENCE MARCH 2019  
PERIPHERAL NERVE SOCIETY  
MEETING JUNE 2019**

**Conclusion:** The substitution effect is a robust entity. Over three-quarters of Chronic Pain (CP) patients receiving opioids reduced their use and two-thirds of patients receiving anti-anxiety, migraine, and sleep medication decreased their use.

The biggest concern with Medical Cannabis (MC) may be economic. MC stigma may be an impediment to full-communication with health care providers.

Although data collection is ongoing, the self-reported efficacy and limited adverse-effects for MC among CMT is promising.

**[CLICK HERE TO VIEW POSTER](#)**

[www.hnf-cure.org/wp-content/uploads/2019/08/EPA\\_MA\\_Poster.pdf](http://www.hnf-cure.org/wp-content/uploads/2019/08/EPA_MA_Poster.pdf)



Lucia Notterpek,  
Allison Moore,  
Estela Lugo

## PNS, It's A Global Effort

Where do the top CMT researchers in the world unite to share leading-edge research? The Peripheral Nerve Society (PNS) meeting! This meeting takes place once a year in cities around the world. This year's location was the stunning city of Genova, Italy, June 22-26 at the Centro Congressi, Porto Antico Di Genova.

There is no better opportunity to connect one-on-one with international experts from multiple disciplines on the topics of Charcot-Marie-Tooth and related Inherited Neuropathies, Inflammatory Neuropathies, pain and more. The four-day program was packed with talks, presentations, collaborative meetings, educational break-outs and poster sessions. HNF was honored to present at the annual Inherited Neuropathy Consortium (INC) meeting on our new Movement is Medicine™ program, an innovative research initiative to bring exercise modalities to all patients with CMT. These studies have the potential to be an adjunct therapy to improve functional outcomes and endpoints in future CMT patient clinical trials.

In addition, HNF is a key partner of the INC and is on the internal Diversity Core Committee to help recruit a diverse patient population for the INC research studies. We are very excited to have implemented a Voice Survey in Spanish to support the Latino community to participate in clinical trials. We launched the study on September 16th and welcome participation!

**TO PARTICIPATE:** <https://www.hnf-cure.org/registry>

Allison Moore, founder/CEO of HNF presented on three research studies spotlighting the Global Registry for Inherited Neuropathy:

**1.**

**CMT1A and Impaired Patient Mobility: Expressions, Remedies and Impact on Quality of Life**

**2.**

**The Path To Diagnosing Charcot-Marie-Tooth Disease: The Patient Experience**

**3.**

**“Examination of Risks/Benefit Profile of Medical Cannabis In CMT and HNPP and Chronic Pain Patients”**

– Brian Piper, Phd.

HNF was front and center at the Pharnext Presentation, as Scientific Advisory Board Member Michael Sereda presented the biomarker results of PXT3003 and world renowned Induced Pluripotent Stem Cells (iPSC) lines researcher Mario Saporta gave a thorough overview of CMT. They highlighted results from pivotal phase III trials for the first potential drug to treat CMT1A, PXT3003.

**“I have waited years for this moment and am grateful to Pharnext for choosing CMT as their first pipeline class of drugs!”**

– Allison Moore, HNF Founder & CEO

A focus group was also hosted by Vitacces in collaboration with Pharnext Pharmaceuticals, for their CMT research app, CMT&Me. HNF was honored to join Mark Larkin, founder of Vitacces and his team, the distinguished advisory board, during this collaborative discussion along with other advocacy groups from around the world: CMTA, CMT France/Europe, CMT UK, and ACMT - Rete (Italy) to ensure that patient voices are being heard and continue to align with the most vital needs of the community.

[To join CMT&ME: Search your app carrier](#)



Allison Moore, CEO, HNF  
Estela Lugo, Medical Outreach Manager, HNF





# CMT Spanish Survey



**CMT Encuesta de voz = 7 minutos**

Comparte tu VOZ con Charcot-Marie-Tooth investigación

TRUE REPLY  
Be heard

Por Favor

Tómese unos minutos para ayudar a Charcot-Marie-Tooth a investigar participando en nuestra Encuesta sobre la Diversidad Global. Es importante que representemos todas las voces de Charcot-Marie-Tooth por igual.

**1-844-960-0914**

HEREDITARY NEUROPATHY FOUNDATION

Part of the Hereditary Neuropathy Foundation's mission is to represent the voices of all CMT patients equally, regardless of ethnicity, nationality, or socio-economic status.

This is why we are initiating our first ever, ground breaking "Global Diversity Voice Survey" using state of the art Voice Activation Technology (VAT) developed by [TrueReply.com](https://www.TrueReply.com).

By capturing the patient experience with CMT in their own voice and native language, HNF hopes to improve access and participation for underserved minority populations in clinical research projects, and to gain deeper insights into these populations particular experience with CMT.

Our first Global Diversity Voice Survey will engage Spanish speaking patients with 9 questions. Participants will be able to answer the questions freely in their own voice; their responses will be recorded, transcribed and translated to English for analysis and insights.



CMT & ME app  
Download available on  
Android or iOS device  
at the app store.



Dr. Lorenzo A. Woodson

## Diversity Inclusiveness and Clinical Trials

Those of us coping daily with Charcot Marie Tooth (CMT) neurological condition know it reaches far and wide across all cultures. I am of African descent, and I am the face of CMT. I am reaching out to all African Americans struggling with the symptoms of CMT who may have given up hope and feel that there is nothing that can be done. Well, there is hope through clinical research studies that are being advanced, and we need your support and participation. We believe here at Hereditary Neuropathy Foundation (HNF) that we can help all cultures by being diverse in our outreach to overcome CMT. If you are of African descent and you are having weakness in your extremities (hands and feet), drop foot, numbness in legs, hands and feet, difficulty running or walking or standing, just to name a few symptoms, please see a medical professional and request to be evaluated for symptoms of CMT. Then feel free to connect with the Hereditary Neuropathy Foundation to begin your journey to helping us find a cure. We welcome you to the CMT family and with your involvement we are becoming more culturally diverse in our outreach for the treatment and eventual cure for the CMT population.

Dr. Lorenzo A. Woodson – CMT Patient

[RECENT INTERVIEW WITH FROM DR. WOODSON. CLICK HERE](#)



## Have you downloaded the CMT&Me app?

Be part of the first groundbreaking research app for CMT! The CMT&Me app collects real-world data using 'bring your own device' (BYOD) technology – participants use their own smartphones to complete questionnaires or surveys at their convenience. The app immediately submits the information patients provide to a central database. A Scientific Advisory Board oversees the CMT&Me study, and includes clinicians who care for CMT patients, Patient Advocacy Organization (PAO) representatives, and experts in patient-reported outcomes (PRO) and data management. Non-commercial researchers and PAOs can apply to the Scientific Advisory Board for access to aggregated data via dashboards.

**Researchers can then use that data to better understand CMT and improve patient care.** All patient data remains anonymous, and researchers who access the CMT&Me study database will not be able to identify patients.

## Available now in Spanish!

Download available on Android or iOS device at the app store.



## Meet HNF's Newest Team CMT member, Lisa Mormann

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I am 46 years old from Iowa and I'm married without children. I spend most of my free time on a bike outdoors or spinning on my smart trainer at home; otherwise I can be found at my local gym.

I've worked mainly in the dental field for the past 30 years. I started working in the back with doctors and patients, which is still where my heart is. However, due to lack of sensation in my hands and inability to be on my feet for eight hours a day, I ultimately accepted a desk position in the same office.

I motivate others not to give up; someone needs to be last. I was that person who never thought she could be part of any sporting event due to my Charcot-Marie-Tooth (CMT). I did not walk as a child, then used braces to walk until I started having reconstructive foot surgeries in my 20s. In 2001, when I went to see my neurologist as a follow up, I was advised to start losing weight and to become more active. At that time, he recommended cycling. I got home and went right to the bike shop and purchased my first bike.

When I was younger, I was depressed that I was not able to play sports and be active like my friends. What was wrong with me? I would fall for no reason, and it was normal to have a sprained ankle. I wrapped myself in a blanket until it reached about 80 degrees and always felt tired. Normal tasks, such as opening a bottle, picking up a pencil and writing, or towelling off post shower are daily challenges. What is more, the Iowa winters really play havoc with me, as I get stabbing jolts, bug-crawling pain, and my feet turn bluish and I don't want to get out of bed. To this day, I don't like to ask for assistance with daily tasks, but sometimes I have no choice. One of my ongoing challenges is accepting that my body requires more sleep and increased recovery time than others.

I did not know I had a Hereditary Sensory Motor Neuropathy



my fellow cyclists know that I may need assistance removing my helmet, unzipping my jacket, and removing my boots, due to lack of sensation in my hands.

I have served on the Dubuque Bike Club board for the past six years. I also started and lead the Tuesday Night Gravel group. My motto for the group is: "The speed of this group is the speed of the slowest rider".

As friends say, “Lisa sets goals, registers for races and puts more mileage on her bicycle than we have ever witnessed from anyone! In fact, her bicycle mileage outnumbers the miles on my car per month”.

The reason I wanted to join the CMT Team is to continue to raise awareness about CMT. Just because I have CMT doesn't mean it has me. I just have to focus and work harder and dig deeper for my goals.

Most of my fellow cyclists know that I fight challenges, and are always there for full support. If I have a bad day on a ride, one of them will stay with me for support and motivation, versus allowing me not to ride. My left leg is 'along for the ride' and my right leg is my 'powerhouse.' However, when I tell other cyclists for the first time about my challenges, they compliment me on my motivation to fight. When I'm out cycling in cooler temps,

## Challenge your limits

My Instagram handle are both: cycling\_chic73  
Facebook: Lisa Mormann





# Swimming Towards Success, Jamal Hill

At a young age, Jamal Hill found his love for swimming through a local YMCA “Mommy & Me” swim class. The Los Angeles native was a natural born swimmer, and has continued his aquatics career throughout his life.

In elementary school, Hill began to swim competitively until he suffered a shoulder injury in 5th grade. Once his shoulder healed, Jamal began training with Conrad Cooper, a local community swim instructor who encouraged his talent. At 10 years old, Jamal was diagnosed with Charcot-Marie-Tooth after falling victim to a bout with paralysis. After Hill’s recovery, his parents encouraged him to never be discouraged by his impairment, and instead to use it as an opportunity to overcome challenges and inspire others.

## Adolescence

Hill attended Junipero Serra Catholic High School in Gardena, California. During his high school career, he swam on the varsity swim team while being an honors student. He developed an interest in medicine which influenced his decision to major in Physics and minor in French at a college in Ohio. After three years of enhancing his craft, Hill left Ohio for the University of Southern California (USC) under the tutelage of Coach Dave Salo, to continue working out with the elite Trojan swim team, which he did through May 2017.

The discipline and determination of Hill helped him maximize the opportunity Coach Salo offered him. He used the training ground to challenge himself, enhance his skills, self-reflect, and network with other swimmers. While attending a UCLA

swim meet, Hill noticed one person consistently swam better than him; that person was Luke Pechmann. The two swimmers became fast friends and, Pechmann introduced Hill to his coach, Wilma Wong, who subsequently agreed to take him under her wing. Under the coaching of Wong, Hill has concentrated tremendously on his technique while improving his ranking over a one-year period from unranked-amateur, to Ranked #1 in the Entire Nation.

## Current Journey

Since 2014, Hill has worked as a lifeguard for the County of Los Angeles. He recently began an initiative to teach One-Million people to swim through private lessons, ad partnerships with swim schools worldwide, in an attempt to lower the global drowning rate. This fall, he will test for certification as a Los Angeles County Fire Department Ocean Lifeguard. The certification also carries a credential as a licensed paramedic.

These licenses and certifications are all part of his preparation for the 2020 Paralympics in Japan, where he will compete in the men’s 4×100-meter freestyle relay, the 4×100-meter medley relay, the mixed 4×50 meter relay, the men’s 50-meter freestyle race, and the men’s 100-meter backstroke race. His success in Japan will move him closer to his ultimate goal – the 2024 Paralympics in Paris, France.



# ASK THE EXPERT



## Cannabis & CMT Webinar Overview

CMT-Connect hosted its first webinar on the popular topic of Cannabis this past April with a panel of experts and CMT patients. Brian Piper, PhD and Meg D'elia provided industry and scientific insight with recent studies showing positive results on the effects of CBD and Cannabis on chronic pain and other CMT-related symptoms. CMT patients Bernadette Scarduzio and Di Billick shed light on treating their own pain, fatigue and muscle spasms with different strain types and methods.

“We must do our part to break stigmas around Cannabis in the CMT community and share the most recent validating research. Our community is experiencing extreme and debilitating pain. They deserve safe and effective options”

– Estela Lugo, HNF Medical Outreach Manager

**CLICK HERE TO WATCH THE FULL WEBINAR:**

<https://www.youtube.com/watch?v=fMvwbKzZf5k>

## UPCOMING WEBINARS

How to Exercise in the Pool: September 19, 2019

**REGISTER FOR UPCOMING WEBINARS:**

[www.hnf-cure.org/cmt-connect](http://www.hnf-cure.org/cmt-connect)

## PAST WEBINARS

- |   |  |
|---|--|
| * Ability360 Sports and Fitness Center, with Christina Chambers | * Panetta Physical Therapy                               |
| * Mental Health & CMT   | * CMT & Balance  |
| * CMT & Microcirculation  | * CMT & Your Nutrition                                   |
| * Jamal Hill ~ Paralympic Swimmer                               | * CMT&Me App   |
| * Made for Motus  | * CMT & Finances   |
| * Mobi Mats August 22, 2019                                     | * Ability360 Sports & Fitness Center with Brielle Carter |
| * CMT & Surgery 101 with Dr. Glenn Pfeffer                      | * Active Hands   |
| * Bemer Technology  | * Cannabis & CBD for CMT                                 |
|   | * CMT & Canine Companions                                |

**VIEW PAST WEBINARS:**

[www.hnf-cure.org/cmt-connect-webinars](http://www.hnf-cure.org/cmt-connect-webinars)

Do you ever wish you could have direct access to a Neurologist for your CMT questions? Now you can! HNF is proud to present our new web page featuring real questions from CMT patients across many topics. Submit your questions directly from our site to Dr. Florian Thomas and Dr. Jafar Kafaie today!

**Q: Is CMT linked to brain fog?**

**A: Dr. Florian Thomas:**

Given that CMT in the overwhelming majority of patients is limited to peripheral nerves, there is little biological basis for “brain fog”. However, in addition to intrinsic brain conditions, symptoms such as mental fatigue can result from sleep deprivation and depression which could occur for a number of reasons in people with CMT. Thyroid disorders and vitamin B12 deficiencies can cause “brain fog” and those can be identified with simple blood tests. Furthermore, “brain fog” is common in people who take many medications that affect the brain, e.g. for neuropathic pain, anxiety, depression, insomnia, high blood pressure, and many others.

► **Have a question?**

<https://www.hnf-cure.org/ask-the-expert>



*Movement is Medicine*<sup>TM</sup>



# TOP 10 Reasons to Attend the Movement is Medicine Summit

November 8-9, 2019, Phoenix, Arizona

1. Network and meet new peers in the CMT community
2. Choose from over 30 interactive & adaptive sessions
3. Learn practical tips and strategies to stay active
4. Share your best practices and lessons learned
5. Hear experts and dynamic speakers
6. Try something NEW in a safe environment
7. Network with CMT researchers and industry
8. Learn to be a better advocate for yourself
9. Build self-esteem and confidence
10. Connect and share your story with others



## HNF AND INSPIRE PRESENT POSTER AT GLOBAL GENES 2019 RARE DRUG DEVELOPMENT SYMPOSIUM

In June 2019, Allison Moore, Founder/CEO, HNF and Judy Chandler, Director of Partnerships, Inspire, presented the poster “Partnership and progress for a rare disease community: How Inspire and the HNF collaborated to grow the CMT patient community and accelerate research”

In 2009, HNF partnered with Inspire, a social network for health, to launch a CMT online support community. The community has grown to more than 6,000 patient and caregiver members and generated 40,000+ posts – real-world data that reveal important trends, themes, and gaps. The poster illustrates how the partnership between HNF and Inspire contributed to several research and educational initiatives:

### Incorporating Pain as a Novel Research and Clinical Outcome Measure

Using pain-related conversations and resulting data from the CMT support community, HNF developed initiatives to understand the potential link between pain and CMT, including a patient-centered pain summit and a patient-reported IRB outcomes study through their patient registry, Global Registry for Inherited Neuropathies (GRIN). Pain is now a clinically relevant endpoint in CMT treatment and key outcome measure in CMT clinical trials.

### Understanding Psycho-Social Components of CMT

HNF leveraged data and content from the CMT support community about the psycho-social components of the disease to develop questions for a Natural History Study and to plan an FDA Externally-led Patient Focused Drug Development (EL-PFDD) Meeting and a Patient-Centered Behavioral Health Summit.

### Capturing the Patient Voice

HNF and Inspire collaborated to use voice-activated technology to capture the CMT patient experience. Results were shared at the EL-PFDD Meeting to help HNF describe the true burden of living with CMT and how patients view the risks and benefits of CMT treatments to inform the FDA, drug developers, and other key stakeholders.

### Developed CMT Healthcare Provider Directory

In 2016, Inspire collaborated with HNF to design and analyze a healthcare provider (HCP) survey in the CMT support community. The survey received 225 responses and resulted in a national directory now over 1,500 providers across many disciplines who understand how to manage patients with CMT.

Inspire and HNF were uniquely positioned to create a successful, innovative partnership beyond the critical work of peer-to-peer support for patients and families affected by CMT.

**JOIN HNF'S INSPIRE COMMUNITY TODAY!** <https://www.inspire.com/groups/charcot-marie-tooth-cmt/>

**CLICK TO VIEW THE POSTER:** [www.hnf-cure.org/wp-content/uploads/2019/09/HNF-Inspire-posters.pdf](http://www.hnf-cure.org/wp-content/uploads/2019/09/HNF-Inspire-posters.pdf)

Dear Lainie,

I just got my first pair of afos and I'm having a hard time with it. I have always been fairly confident and the type of person who doesn't care what others think, but I'm feeling conflicted on whether or not to show my braces to the world, or to keep them concealed? When I have worn them out & about, I've felt very self-conscious and not like myself. What should I do?

From,  
Mara P.

Dear Mara,

Thank you for being real and sharing your feelings. Many leg brace wearers, including myself, can relate. First of all, there are no “shoulds” or rules when it comes to whether or not to wear afos on the outside of your clothing or not, the decision is completely up to you. I promise that you will not be kicked out of the afo wearer's club either way. Having a disability and wearing leg braces are either a big part of your identity or they are not.

As an example, I have two friends who are 10+ years breast cancer survivors. One of them chooses to be an advocate, while the other friend does not identify herself in any way with being a cancer survivor.

While many afo wearers choose to “show their braces” and feel confident doing so, there are just as many of us (including myself) who choose to wear clothing over our braces and to conceal them. The most important thing is how you feel about yourself and your appearance when out & about in the world. It's human nature to care what others think, and first impressions are in part, based on physical appearance. If you don't feel your best showing them, then don't. If in a month or 5 years you decide to rock your braces on the outside of your clothes, then by all means, go for it!

**FOR A FREE AFO GUIDE CLICK HERE.**



Xoxo  
Lainie

**Lainie Ishbia, MSW**  
Disability Speaker & Blogger  
Trend-ABLE





Above left:

Grace's Courage Crusade  
Tonia Hassel, Marybeth Caldarone,  
Carol Liu

## HNF EVENTS recap

### Grace's Courage Crusade Last Bash

On July 21, 2019 at the Charlestown Rathskeller Tavern in Charlestown, RI, CMT 2A supporters came together one last time to support the Caldarone/Sidoti families to fund research for CMT2A. The event was an afternoon of fun for all. The buffet lunch was fantastic, topped off by an ice cream bar that kept everyone energized for the exciting cornhole tournament. Local businesses and supporters donated baskets and individual prizes, which kept participants on the edge of their seats for the raffle that closed out the evening.

We thank everyone who attended.  
**We raised more than \$16,000 for CMT2A research!**

**"HNF is grateful for the support of the Caladrone & Sidoti families as they help fund research for CMT2A."**

—Courtney Hollett, Executive Director, HNF

### James Cure raises over \$72,000

On Friday, July 26, the McCarthy's hosted our first golf fundraiser for our son James and other children who have CNTnap1, James' Scramble For a Cure, at Tijeras Creek Golf Club in Rancho Santa Margarita, CA. One hundred golfers came out in support of the family, despite the heat and humidity! Golfers came ready to play, but were first served lunch by a local pizza restaurant, Selma's, which served up pizza and beer to all who attended. After the golf round, an additional 100 guests arrived for the dinner festivities, which included a silent and live auction. Guests were immediately drawn to our helicopter golf ball drop raffle, which was fun for all! A helicopter dropped almost 700 golf balls that were sold for a chance at prize money. We all watched on the patio as the golf balls came flying towards the hole and couldn't wait to find out who the winner was! The first ball to go in to the hole was determined the winner which belonged to a local charitable

group called Generation Give. They are truly incredible, and elected to give back their winnings to the Hereditary Neuropathy Foundation! Thank you, Generation Give. We appreciate your support and generosity.

From left to right:

Judy Pratt, Erin Allen, Brad Bermingham, Eddy Montoya







**Above:** James Cure golf fundraiser

The night was finished off with a tasty dinner, a silent auction with over 70 items, ranging from wine tastings at a winery to signed sports memorabilia, and also a live auction. A great time was had by all of the guests, and we couldn't be happier with the results of our first event and the support our family received. It's amazing how much love and generosity our family felt that night and we are in awe of our community. Thank you to the HNF team who was extremely supportive and helped make the event such a success! With the help of HNF and our event planning team, Twenty156, we are confident that we made our goals that we set for fundraising and are looking forward to finding out more about future treatment options for our son.

Thank you to all of our sponsors, Art Store Fronts, Redwood West, AMA Consulting Engineers, hole sponsors

and guests for their support, we couldn't have had such a successful first tournament without you.

Thanks,  
Kyle and Lindsey

## An Evening for Charity Take Two

On Saturday, April 6th, HNF hosted its annual Evening for Charity event at CoolMess in Roslyn, New York. The night was filled with food, drinks, music, auction items and casino tables. Guests mingled and enjoyed dining and dancing, while admiring and bidding on numerous silent auction items.

Krystyna J, said, "It was a special night for a special cause. The gathering of

people had such fun gambling, mingling and eating good food knowing that they were helping to find a cure for CMT."

Thanks to all who joined us, this special evening raised over \$25,000 for HNF's programs! We were thrilled to see so many people with CMT (old friends and new ones), family members, and friends coming out to support HNF. We are grateful for their generosity.

A special "thank-you" goes out to the Loucas', Dellis', and Cyprus' family, owners of CoolMess. Marguerite Loucas, restaurateur, is a childhood friend of HNF CEO Allison Moore and a founding board member of HNF.

Marguerite noted, "It was an honor and a privilege to host this great event at CoolMess. Allison and the team at HNF are doing amazing work to help find a cure for CMT disease."

**It's not too late to donate:**  
<https://curecmt.givesmart.com>



**Above:**  
An Evening for Charity

**Left:**  
John Panico, Irene Dellis, Gregory Dellis

**Far Left:**  
Tom & Anna Cahill, Matt & Donna Downing



# 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**  
Los Angeles, CA  
**Contact:** Dana Fine  
310-423-8497  
Dana.Fine@cshs.org

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

## CONNECTICUT

**Hospital for Special Care**  
New Britain, CT  
**Contact:** Sharon McDermott  
860-612-6305

## FLORIDA

**AdventHealth Neurology at Winter Park**  
**Contact:** Nivedita Jerath, MD, MS  
407-303-6729  
nivedita.jerath@gmail.com

**University of Florida Health**  
Gainesville, FL  
**Contact:** Tracie Kurtz, RN, CCRP  
352-273-8517  
tlkurtz@ufl.edu

## University of Miami

Miami, FL  
**Contact:** Meri Jaime (for appointments)  
305-243-7400  
MJaime@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:** Nicole Jenci  
913-945-9934  
njenci@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:** Susan Eller  
314-977-4867  
ellersc@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:** Annerys Santos  
551-996-8100  
Annerys.Santos@HackensackMeridian.org

## Atlantic Health System\*

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

## NEW YORK

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

## WASHINGTON

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

\* Pediatric Center of Excellence

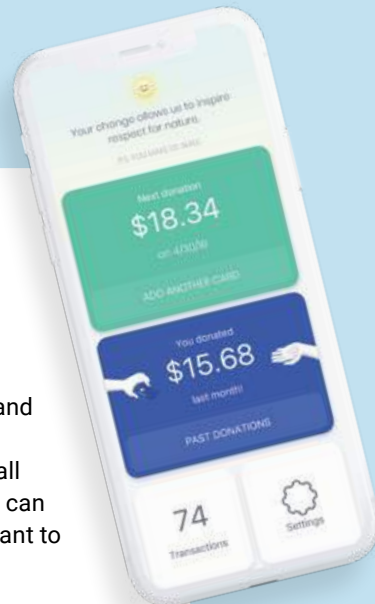
## Help Support HNF with the RoundUp app!

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](http://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](mailto:courtney@hnf-cure.org)

Thanks,  
Courtney



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# Attention!

## Team CMT Event Opportunities

HNF is proud to be a charitable partner for the following Team CMT event in 2019. All funds raised will go to HNF's TRIAD program for CMT research.

### **TCS New York City Marathon** SUNDAY, NOVEMBER 3<sup>RD</sup> New York City

We are thrilled to be headed back for the second time to the TCS NYC Marathon. The annual marathon course takes the runners through the five boroughs of New York City. In 2018, the race had more than 52,000 finishers, the largest field event in history. Team CMT participated in the 26.2 mile race through the Big Apple last year and celebrated as HNF's Founder/CEO, Allison Moore crossed the finish line with her friend and HNF Chairman, Joy Kaye.

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To join HNF's Team CMT at any of these events, please email [courtney@hnf-cure.org](mailto:courtney@hnf-cure.org)

## New Board Member



**Robert A. Kaufman** is the President of Healthcare Risk Advisors, formerly FOJP, and Hospital Insurance Company, a metropolitan New York based insurance company that provides comprehensive insurance and risk management advisory services to a group of major hospitals, long-term care facilities, and social service agencies.

Rob has built a distinguished legal career as a strategic advisor on corporate governance, insurance regulatory issues, compliance and risk management. Prior to joining FOJP in January 2014, Rob was Senior Vice President, Secretary, General Counsel and Chief Compliance Officer at Harleysville Insurance. Rob was also a partner at Reed Smith, an international law firm specializing in complex litigation, strategic transactions, and regulatory matters.

In addition to his private sector experience, Rob served with distinction as an Assistant U.S. Attorney in the Criminal and Asset Forfeiture Divisions of the United States Attorney's Office for the Eastern District of Pennsylvania.

Rob earned his BA and JD from the University of Pennsylvania. He currently lives with his family in New York City and has CMT1A.



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