HEREDITARY NEUROPATHY FOUNDATION

WINTER 2022

cmtupdate

CMT UPDATE
WINTER 2022

Allison T. Moore
Founder and CEO
Hereditary Neuropathy Foundation

I am so excited to invite you all to join us virtually at HNF's Patient-Centered CMT THRIVE Summit™ on March 5-6, 2022. Why THRIVE? The definition of THRIVE is "grow or develop well" or "prosper; **flourish**". After two years of living with the challenges of COVID-19, in addition to the daily challenges of living with CMT, we are ready to THRIVE. I'm very proud of the agenda and speakers that have been curated for you by the amazingly talented Estela Lugo, HNF Project Development Manager! Our aim is to uplift the CMT Community in all areas of their lives: our mission is to have attendees come away with a new and brighter perspective on disability and be empowered with many tools to help navigate their journeys toward a more fulfilling life with CMT.

In addition to supporting people living with CMT and their families with our Patient-Centered CMT Summits™, HNF is investing in research and development of treatments and potential cures for all types of CMT. Read on to learn about our three new drug development projects, our new clinical research mobile app, three new gene therapy development projects, and five clinical trial development projects. We have been very busy! All of these projects require funding and, as always, we are grateful for your generous support and continued diligence in spreading the word about supporting HNF through your local fundraising efforts, social media campaigns and matching funding through your employers. We are proud to announce that we met the $200,000 matching challenge for CMT2A research! If you would like to establish a matching challenge for your type of CMT, I'd love to hear from you.

Many of our industry partners are thriving as well! Be sure to read about the on-going recruitment for the Pharnext PREMIER trial. We also want to give a big shout out to two of our amazing athletes of Team CMT: Brad Floyd, who ran the 2021 TCS New York City Marathon to support his spunky daughter, Addie, and Kat Guenther who just completed two half marathons. I hope you'll consider joining Team CMT or our Movement is Medicine™ weekly classes to just keep moving and thriving in 2022.

I hope you'll enjoy this edition of HNF's *CMTUpdate*, and I look forward to hearing from you to hear how you THRIVE with CMT.

Best,

Allison T. Moore

THRIVE

SAT & SUN, MARCH 5-6TH 2022

Join us on March 5-6th for a weekend of THRIVING!
HNF is thrilled to invite our CMT families and friends to our 2022 Patient-Centered Charcot-Marie-Tooth **THRIVE Summit!**

THRIVE

For 2 days, HNF's stellar line-up of speakers and sessions will aim to uplift the CMT community in ALL areas of their lives!

Sessions will include hand-selected speakers who are THRIVING with disabilities & can speak from first-hand expertise and experience on their subject matter.

Our Goal is to have attendees come away with a new and brighter perspective on disability and empowered with a plethora of tools to help navigate their journeys towards a more fulfilling life with CMT.

Session Categories:

- Self Love & Advocacy
- Family & Relationships
- Work & Career
- CMT Research
- Kids + School + Social
- Accessibility

***NEW* Speed Networking** (optional) will allow attendees to randomly match with each other on screen for a 3-minute CMT meet & greet! Choose to chat longer or match to the next attendee!

Private Virtual Consultations for Surgery & Genetic Testing

AGENDA

Day #1 - Saturday, March 5

SELF LOVE & ADVOCACY

Welcome "Thrive" Meditation
Presented by: Shakti Bell

Shakti is a certified Yoga Therapist living with MS. During this guided meditation, Shakti will lay the foundation for opening ourselves to self-love and new possibilities.

"How to Talk to Anyone When You Have A Disability"
Presented by: Lainie Ishbia, Trend-able & Estela Lugo, Hereditary Neuropathy Foundation

Lainie & Estela live with CMT are firm believers that the key to THRIVING starts with how we feel about ourselves. This interactive workshop combines roleplaying, communication hacks, humor, and vulnerability for a fun-filled experience bound to boost anyone's self-advocacy skills and daily interactions.

FAMILY + RELATIONSHIPS

The Impact of Healthy Relationships
Presented by: Rachelle Friedman, LCSW EMDR, ART, Brainspotting, EFT Therapist, Brainy Social Worker

It's been said that the five people closest to us shape our success, our interests and much more than we even realize. If the people we surround ourselves with affect us on such a fundamental and human level, it's absolutely vital to learn how to attract those we admire while phasing out toxic interactions.

Dating + Love + CMT
Presented by: Shonna Counter

Dating, relationships, and intimacy play significant roles in the human experience. We understand the challenges many of us can encounter when CMT is added to the mix.

Caring for CMT Caregivers
Presented by: Laura Russell, MSW, LCSW Neuromuscular Medicine – Social Worker, AdventHealth

We can't stop the impact of CMT on someone for whom we care. But there is a great deal we can do to take responsibility for our well-being. Gain new tools, skills, and resources for navigating life as a caregiver during this session.

WORK / CAREER

Working as a Disabled Executive
Presented by: Sheryl Markowitz, MSW, LCSW, LISW-S

As the senior executive of Healthfirst, Sheryl shares her personal story on navigating the corporate world while living with CMT. Her empowering tips for addressing the physical and emotional challenges redefine what success means to both herself and the workforce at large.

Connecting with Inclusive Employers – Inclusively Interview & Demo
Presented by: Sarah Bernard, Inclusively COO

Inclusively is an employment platform for job seekers with disabilities and goes beyond a candidate's background, matching their experience, skills, and accommodations needed to succeed in the role. Inclusively COO, Sarah Bernard will share how candidates can connect with their ideal employers while honoring their required accommodations during this session.

Know Your Disability Rights 101
Presented by: Ashley Jacobson Esq., MA, CRC, Jacobson Law and Advocacy

What are our disability rights at work, school & society in general? When it comes to CMT, learning our rights can seem like a daunting and intimidating endeavor. Thankfully, there are passionate and empathetic Disability attorneys like Ashley Jacobson that help us navigate equality and open the doors to inclusion!

Applying for Social Security Disability 101
Presented by: Nancy Cavey, The Law Offices of Nancy L. Cavey

The Social Security Administration (SSA) doesn't make it easy for those with CMT to get the Social Security Disability Benefits they deserve. SSA is denying more initial claims and Requests for Reconsideration claims than ever for CMT. That's why attorneys like Nancy Cavey work with applicants to provide important guidelines to ensure no details slip through the cracks during the SSA's 5 Step Sequential Evaluation process.

(Agenda continued on next page)

Day #2 - Sunday, March 6, 2021

CMT RESEARCH

The Future of CMT Research

Presented by: Allison Moore, Founder and CEO, Hereditary Neuropathy Foundation

An Overview of HNF's Therapeutic Research in Accelerated Discovery (TRIAD) projects and latest research milestones.

Can Existing Drugs Treat CMT?

Presented by: Omid Karkouti, Co-Founder and COO, Rarebase

HNF, in partnership with Rarebase, is leading the charge in the first-ever research initiative to tackle multiple types of CMT in one project using its tech-enabled drug discovery platform called "Function." Rarebase will screen a compound library of thousands of FDA-approved drugs and novel drugs, targeting ten subtypes of CMT. Join Rarebase, Founder & COO Omid as we dive deeper into this exciting new technology and what it means for treating CMT.

The Key Role CMT Patients Play in Research

Presented by: Joy Aldrich, GRIN Patient Registry Coordinator, Hereditary Neuropathy Foundation

Without your participation, researchers won't have the essential patient information to develop drugs, gene therapies, and clinical trials for Charcot-Marie-Tooth and other Inherited Neuropathies. In addition, as GRIN grows, we gain greater insights from you as patients to help accelerate therapies for Charcot-Marie-Tooth (CMT) and Inherited Neuropathies.

Cannabis & Neuropathy: a focus on improving patient quality of life

Presented by: Miyabe Shields, PhD & Andrew Westercamp

Cannabis can be a safe and effective tool to reduce symptoms of neuropathy and improve patient quality of life. The first part of this presentation is a brief overview of how cannabis interacts with the human body to reduce neuropathic pain by a scientist who specializes in cannabinoid pharmacology. The second part provides patients with tangible takeaways for how to use different cannabis products from a cannabis cultivator and industry entrepreneur with HNPP. Real Isolates LLC is a cannabis research startup created for patients by patients.

KIDS - SCHOOL & SOCIAL

Empowering Kids with CMT

Presented by: Dr. Gabrielle Ficchi, Founder and CEO, New Perspectives, LLC

What are the key ingredients and practices for raising empowered kids with CMT? Dr. Ficchi is a licensed therapist with specialty areas of focus on individuals with disabilities. In addition, her personal experience with a disability (Cerebral Palsy) helps her relate to participants' experiences. Gabrielle's work centers on the family experience including how parenting styles influence social maturity in children and adolescents with physical disabilities and influence their transition to adulthood.

School & CMT – "What I wish I knew sooner"

Presented by: Brooke Warren, Parent & Advocate

How do parents support their children while navigating the school system with CMT? What are the best practices for advocating for their needs, communicating with educators, requesting services, and helping our kids THRIVE inside the classroom. Join Brooke Warren for an insightful presentation & discussion on important lessons learned from her personal experience as the parent of a child with CMT.

Top Therapies for Kids with CMT

Presented by: Nivedita Jerath, MD, MS, Neuromuscular Neurologist AdventHealth

What are the key CMT milestones, markers, and interventional therapies to consider when raising a child with CMT? Dr. Jerath breaks down the most beneficial pediatric treatments used in her practice at the CMT Center of Excellence.

Show & Tell: CMT Kid Hacks

Families show & tell their everyday CMT hacks with videos and photos of their creative workarounds for living with CMT.

(Agenda continued on next page)

AGENDA (CONTINUED)

ACCESSIBILITY

Accessible CMT Home Tour

Presented by: Stephanie Carmody, MS SLP-CCC
Dennis Carmody, MPH

What does an accessible home look like? What types of features, appliances, and upgrades can we add to make life easier and maintain independence at home? Join Stephanie and Dennis Carmody for this fun and CMT-friendly home tour, complete with their favorite spaces, tips and products for Thriving!

CMT-Friendly Fashion – Shoes, Clothing & Accessories

Presented by: Lainie Ishbia, Trend-able.com

How can we style up our closets with AFO-friendly footwear, fashion and accessible finds? Join Lainie Ishbia for a virtual TREND-ABLE shopping tour complete with clever hacks for looking and feeling fabulous! *Styles for all genders*

Adaptive Driving for CMT 101

Presented by: Susie Touchinsky, OTR/L, SCDCM, CDRS, Adaptive Mobility Services, LLC

Driving is vitally important to everyone, but SAFETY is the number one priority. CMT does not require us to stop driving, but it does mean we take the steps necessary to continue driving responsibly and in a way that feels comfortable for everyone. Join us for a crash course on the process of adaptive driving from evaluations, equipment, and certification!

Accessible Travel

Presented by: Wheel the World

Find and book the most amazing accessible places to stay, things to do and multi day trips for people with disabilities.

Wheel the World is an online travel marketplace with the purpose to make the world accessible, one destination at a time.

REGISTER FOR THIS TWO-DAY EVENT

Click Here!

Virtual

HEREDITARY NEUROPATHY FOUNDATION

THRIVE

PATIENT-CENTERED CMT SUMMIT

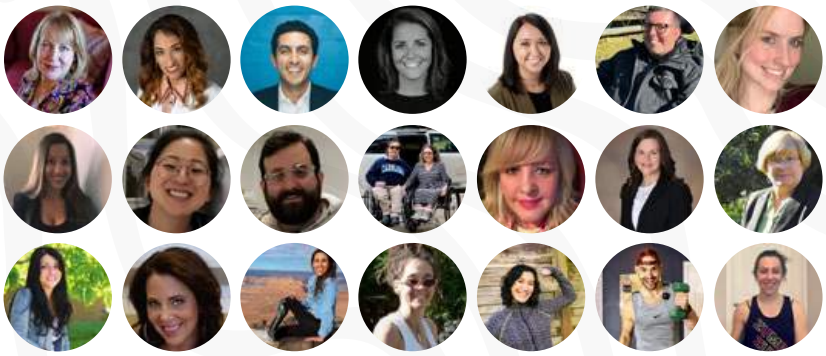
SAT & SUN, MARCH 5-6TH

OVER 25 SESSIONS

SESSION TOPICS:

- SELF LOVE & ADVOCACY
- FAMILY & RELATIONSHIPS
- WORK & CAREER
- CMT RESEARCH
- CANNABIS FOR CMT
- KIDS + SCHOOL + SOCIAL
- ACCESSIBILITY
- ADAPTIVE DRIVING
- ADAPTIVE FASHION
- ACCESSIBLE TRAVEL
- LIVE Q&A & MORE!

OVER 21 GUEST SPEAKERS



OVER 20 BOOTH EXIBITORS

PLUS SURGERY CONSULTATIONS

PLUS SPEED NETWORKING

RESERVE YOUR SEAT!

SPONSORED BY TAYSHA GENE THERAPIES

TIX & AGENDA: **CMTSUMMIT.COM**

TRIAD
THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY

3 New Drug Development Projects

1. RAREBASE – REPURPOSING DRUGS

In partnership with Rarebase, HNF is leading the charge in the first-ever research initiative to tackle multiple types of CMT in one project. Rarebase is a public benefit biotech company focused on accelerating therapy development for rare diseases with its tech-enabled drug discovery platform called "Function". Rarebase will screen a compound library of **thousands of FDA-approved drugs and novel drugs**, targeting 10 mutations of CMT.

- *CMT1A & HNPP (PMP22 dup & del)*
- *CMT2A (MFN2 & MFN2 w/ optic atrophy)*
- *CMT6 (Leigh's Syndrome) (C12orf65)*
- *CNTNAP1*
- *CMT4A & CMT2K (GDAP1)*
- *SORD Deficiency*
- *CMT4J*
- *MTMR2*
- *PRX*
- *OPA1*

Funding needed (Goal = $150k):

HNF aims to develop additional human-derived cellular models for testing. To learn more and support the development for your subtype of CMT, contact allison@hnf-cure.org.

DONATE TO RAREBASE ➜

2. NOVEL DRUG FOR MULTIPLE CMT TYPES

HDAC6 Inhibitors – HNF was the first advocacy group to fund research for the basic understanding of how HDAC inhibition impacts CMT2A by testing this class of drugs in animal models such as zebrafish and mice for validation. Since 2014, HNF has invested $425,000 in HDAC6 inhibitors.

- ***The drug is currently being optimized for human trials and advancing to a pre-IND stage.***
- ***Undisclosed Biotech Company, University of New Zealand, Burke Institute, and University of Sheffield.***

For more on HDAC6 inhibitors, **CLICK HERE** ➤

Funding needed (Goal = $250k):

HNF aims to test HDAC6 in cellular and animal models with other subtypes of **CMT, such as CMT1A, CMT4A, CMT2K, and CMT6**. To learn more and support the development for your subtype of CMT, contact allison@hnf-cure.org.

DONATE TO HDAC6 ➜

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TRIAD
THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY

3. AUTOSOMAL DOMINANT OPTIC ATROPHY ADOA (OPA1)

In partnership with Fondazione per la Ricerca Biomedica Avanzata Onlus – Veneto Institute of Molecular Medicine – V.I.M.M., **HNF awarded a grant to accelerate the screening of FDA approved drugs to identify small molecules that counteract axonal mitochondrial depletion in ADOA Retinal Ganglion Cells (RGC's)**.

- *Autosomal dominant optic atrophy (ADOA)-plus syndrome is a rare inherited neuropathy affecting the OPA1 gene that involves vision loss, weakness in the muscles that control eye movement (progressive external ophthalmoplegia), difficulty with balance and coordination (ataxia), hearing loss, disturbances in the nerves used for muscle movement and sensation (motor and sensory neuropathy), and muscle weakness (myopathy).*
- *HNF expects to have an initial progress report in mid 2022.*

For more information, **CLICK HERE ➜**

1 New Clinical Research Mobile App

CAPTUREPROOF

In partnership with CaptureProof, HNF developed a mobile app as an extension of the Global Registry in Inherited Neuropathies (GRIN) that uses advanced computer vision in a smartphone's live camera to document CMT characteristics, symptoms, and functional challenges with activities of daily living (ADLs).

- *CaptureProof is empowering patients across the US to tell their CMT stories. During this data collection, patients will answer survey questions, take photos and videos specifically designed to allow CMT experts (or Artificial Intelligence) to diagnose and measure the progression of CMT.*
- *Not only is this a safer way to visit with doctors during a pandemic, but it will provide access to CMT experts virtually documenting real-life challenges.* ***With CaptureProof, HNF is putting a CMT Center of Excellence in living rooms across the US!***

To enroll in this study, contact registrycoordinator@hnf-cure.org.

Funding needed (Goal = $500k):

HNF aims to expand the pilot study globally to include multiple languages and countries where specific subtypes of CMT are required for clinical trials. The funds will support CMT Key Opinion Leaders in various countries. For more information, contact allison@hnf-cure.org.

DONATE TO CAPTUREPROOF ➜

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TRIAD
THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY

3 New Gene Therapy Development Projects

1. GDAP1 (CMT4A)

HNF expanded its aggressive mission to develop potential treatments for CMT4A, which includes a viral vector-based replacement gene therapy. In 2019, HNF was the first to recognize this gene's high potential for gene therapy success.

HNF has reached several milestones:

- ***The funding and development of human-derived cellular models by Dr. Mario Saporta at the University of Miami have been successful.***
- ***The funding and development of a rat model in partnership with Envigo, Burke Neurological Institute Academic Affiliate of Weill Cornell Medicine is currently in progress with expected results in early 2022.***
- ***Enrolling patients in a Natural History Study for the development of clinical trials.***

To enroll in the study, contact registrycoordinator@hnf-cure.org.

Funding needed (Goal = $1.5 million):

HNF aims to expand the GDAP1 Natural History Study globally to include multiple languages and countries that provide genetic testing and test viral vectors in cellular and animal models. For more information, contact allison@hnf-cure.org.

DONATE TO GDAP1 ➜

2. C12orf65 (CMT6)

Since 2017, HNF has developed the gene therapy program for CMT6. The viral vector was made, but challenges in models delayed testing. We have pivoted our program to include additional partners hoping that we have overcome the challenges and will be testing the gene therapy in mid-2022.

HNF has reached several milestones:

- ***The funding and development of a viral vector at UNC awaiting optimal cellular and animal models for testing in collaboration with Jackson Laboratory, Helsinki University, and the University of Miami.***
- ***Enrolling patients in a Natural History Study for the development of clinical trials.***

To view update **CLICK HERE** and **HERE ➜**

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TRIAD
THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY

3. CNTNAP1

In November 2018, HNF was introduced to CNTNAP1, a devastating and fatal type of inherited neuropathy. Two families have championed the development of a gene therapy to save these children. In March 2020, HNF initiated a strategy and has met the following milestones.

- *In collaboration with the University of Texas Health Science Center at San Antonio, the development of three mouse models replicating the various mutations within the CNTNAP1 gene. Two of the models have been completed, but unfortunately, time was not on Isabella and her family's side.*

HNF mourns the loss of Isabella, a precious little girl who during her life and afterward, has continued to drive urgent research for this type of CMT. Condolences can be emailed to courtney@hnf-cure.org for her to share with Isabella's Dad (Martin J.).

- *Enrolling patients in a Natural History Study in collaboration with Atlantic Health for the development of clinical trials.*

5 Clinical Trial Development Projects

1. PHARNEXT

HNF has been partnered with Pharnext since 2013 to help identify and facilitate principal investigators & clinical trial sites, protocol development and raise awareness.

- *Pharnext is the first biotech company with an approved FDA IND for clinical trials (Phase II and Phase III) for CMT1A.*
- *The drug PXT3003 is in its final stages for the completion of the Phase III with results expected in Q3 2023.*

*For the latest video update on the PXT3003 clinical trial, **WATCH THE PRESENTATION ➜**

2. APPLIED THERAPEUTICS

HNF has been identifying patients for free genetic and sorbitol screening for potential enrollment in their clinical trial expected to start early 2022. HNF is funding a SORD Deficiency rat to conduct additional studies.

- *This new identified mutation in the SORD gene may be the most common Type 2 CMT. Those patients that are clinically diagnosed with Type 2 CMT may very well have the SORD gene.*

*For the latest on SORD Deficiency, **WATCH OUR WEBINAR ➜**

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TRIAD
THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY

3. ACCELERON

HNF was instrumental in the development of the clinical protocol for ACE-083 to treat CMT1 (the disease's most common subtype) and CMTX (X-linked form of the disease). In collaboration with Acceleron, HNF conducted the largest Patient-Reported IRB-approved research study. The manuscript will be submitted in 2022 for peer-reviewed publication consideration to support future clinical trial designs.

- ***Although the drug failed, the trial gave us additional insights on improving clinical trial protocols, identifying important outcome measures (i.e., pain, fall risks), and what matters most to patients.***

*For more on this study, **CLICK HERE ➜**

4. TAYSHA

In 2014, HNF partnered with Hannah's Hope Fund (HHF), the advocacy group that championed and funded the gene therapy program for GAN. This partnership created the Global Registry for Inherited Neuropathies (GRIN), the leading resource for enrolling GAN & CMT patients into clinical trials.

- ***In 2021, Taysha Gene Therapies acquired exclusive worldwide rights to the clinical stage AAV9 Gene Therapy Program for GAN.***
- ***GAN is known to be fatal in the early onset of the disease; therefore, it's critical that HNF help identify and diagnose patients early for potential enrollment in the clinical trial.***
- ***If successful, this will be the first approved gene therapy for any subtype of CMT.***

5. HELIXMITH

In partnership with Helixmith, HNF is providing critical data from the patient registry, GRIN, and other resources as they prepare for the US trial. This will support the development of baselines for CMT pain, fatigue levels, and other important endpoints.

- ***This will be the first gene therapy phase I human clinical study for CMT1A. The first study was conducted in South Korea with a cohort of 12 patients who had mild to moderate CMT1A.***
- ***The drug Engensis (VM202) was well-tolerated and the safety endpoints were achieved with a trend of improvements seen in the chosen endpoints: FDS, ONLS-leg, CMTNS-v2 (measuring fatigue and neuropathy), and a reduction of muscle loss as evaluated by MRI.***
- ***A larger second study is being planned in the US.***

> *"HNF was the first to acknowledge and conduct research studies with funding support from Patient-Centered Outcomes Research Institute (PCORI), specifically addressing pain in CMT patients. We are thrilled to support Helixmith with our research findings and hope that this will be the first disease modifying treatment that addresses pain."*
>
> – Allison Moore, HNF Founder & CEO

DONATE TO TRIAD ➜

TRIAD
THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY

Donate to CMT2C with A TRPV4 mutation

Click here!

Attention All with CMT2C with a TRPV4 Mutation- Join the New Johns Hopkins Registry!

CONTRIBUTOR: **STEPHANIE CARMODY,** TRPV4 NEUROPATHY PATIENT

Working with the Inherited Neuropathy Consortium (INC), Dr. Brett McCray and Dr. Charlotte Sumber at Johns Hopkins have just established a TRPV4 related Hereditary Neuropathy Registry/ Natural History Study to gain more knowledge about the natural history of TRPV4 neuropathy, learn more about the range of associated symptoms, and to determine if there are ways to detect and measure disease over time. This database is the critical first step that will lay the groundwork for a future clinical trial investigating the effectiveness of a treatment to improve symptoms of the TRPV4 Hereditary Neuropathies (Charcot-Marie-Tooth type 2C, Scapuloperoneal Spinal Muscular Atrophy, and Congenital Distal Spinal Muscular Atrophy). We need as many individuals as possible with TRPV4 HN to join the study! Please email principal investigator Dr. Brett McCray at bmccray3@jhmi.edu or his study coordinator Simone Thomas at sthom125@jhmi.edu to participate. The Johns Hopkins team has also recently established a new Channelopathies clinic for those with TRPV4 related Hereditary Neuropathy to learn more about this condition. This clinic will provide thorough care specific to the needs of individuals with TRPV4 mutations, including interdisciplinary care from neurologists, ENTs, and orthopedists.

To make an appointment in the Channelopathies clinic, **call the Johns Hopkins Outpatient appointment line at 410-614-1196.** If you have any questions, please email Stephanie Carmody at trpv4hngroup@gmail.com.

CLICK HERE! ➜

YOU DID IT!

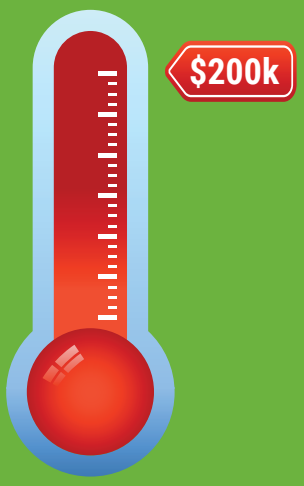
HNF is happy to announce we reached our **matching goal of $200,000 for CMT2A research**. Our Therapeutic Research In Accelerated Discovery (TRIAD) initiative for CMT2A includes many global partners with promising results to enable us to fast-track to clinical trials.

Your precious dollars will support the following pathways:

1. Advance HDAC6 to clinical trials (Undisclosed Biotech)
2. FDA Approved Function Drug Screening (Rarebase)
3. AI Mobile App for Clinical Trial Development (CaptureProof)

The $200,000 will support the TRIAD initiative for CMT2A. Your generosity today will fuel our continued innovation.

Donate to CMT2A ➜



THANK YOU FOR YOUR SUPPORT!

TRIAD
THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY

HNF is proud to announce that Allison Moore will be presenting at the 2022 MDA Clinical & Scientific Conference on March 13 - 16, 2022

The MDA conference will highlight unprecedented research advancements and clinical achievements in Neuromuscular Diseases (NMD), including Charcot-Marie-Tooth (CMT) Disease. HNF shares the same vision as MDA in transforming care through innovation. Allison is super excited to be presenting in the session titled: Latest Developments Across the NMD Registry Data Landscape. She will present the latest developments of HNF's patient registry, Global Registry for Inherited Neuropathies (GRIN) to continue to build strong relationships with advocacy, key influencers and stakeholders. The HNF team will also be exhibiting and they look forward to this unique opportunity to mingle with world leaders and ground-breaking innovators.

The conference will be held at the Gaylord Opryland Resort & Convention Center in Nashville, TN, on March 13 - 16 both in-person and virtual. **TO REGISTER ➜**

TO JOIN GRIN ➜

Meet HNF's Newest TRIAD Council Member Tara Jones, CGC

Tara Jones is a genetic counselor in adult neurology at Cedars-Sinai, with a special focus on neuromuscular and dementia indications. At Cedars, she is the lead research coordinator for the CMT Natural History Study and Sub-studies of the Inherited Neuropathy Consortium and is involved with their Diversity Core. Tara is passionate about bringing equity and inclusion to clinical research and genetic testing and amplifying voices of those with rare disease.

GLOBAL REGISTRY
FOR INHERITED NEUROPATHIES

NEW CMT Mobile App!
– Telemedicine & More

- Tired of doctors not understanding your CMT symptoms and experience?
- What if you could visually share your daily challenges with your doctor through smart photos and videos?
- What if you could be evaluated for surgery or bracing without traveling for hours?
- What if you had telemedicine access to CMT experts and CMT Centers of Excellence?
- What if you could participate in CMT clinical trials from your home?

We're developing an app for that!

Watch our **VIDEO HERE!** ➜

Introducing the CaptureProof CMT mobile app! HNF is bringing Artificial Intelligence to CMT care, with a team of CMT beta-testers.

I'm ready to download & start!

- If you ARE already registered in our CMT Patient Registry (GRIN), click here
- If you are NOT yet registered in our CMT Patient Registry (GRIN), click here and register and complete the Natural History Survey.
- If you are NOT sure, email click here

What can the CaptureProof CMT mobile app do?

Measures & Stores:

- Foot arch & deformities
- Hand tremors & dexterity
- Gait & balance
- Stair climbing
- Challenges with activities of daily living

How can the CaptureProof CMT mobile app help patients?

- Provides real-life data to determine if CMT treatments & trials are actually working!
- Provides real-life video and measurements that can be shared with your doctors!
- Connects you to CMT experts at Centers of Excellence without leaving your home!
- Speeds up CMT clinical trial development in a post-Covid world - no traveling!

Why do I need to register for GRIN before I download CMT Mobile app?

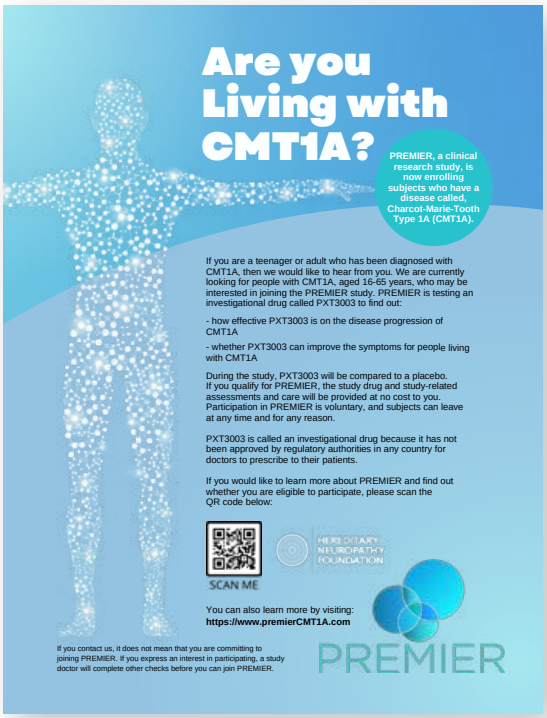
Without your vital (de-identified) medical data, researchers won't have the essential patient information to develop drugs, gene therapies, and clinical trials for Charcot-Marie-Tooth and other Inherited Neuropathies!

Join Movement is Medicine™ Weekly Classes

All classes are offered free of charge

The Movement is Medicine™ mission is to promote the safe adoption of a more active life through exercise and nutrition programs specifically created for those with disabilities.

TO VIEW THE WEEKLY SCHEDULE CLICK HERE! ➜



HNF Partner Pharnext is Actively Recruiting CMT1A Patients for PXT3003 Phase III Trial

Pharnext, a French biopharmaceutical company, is conducting an international study called **PREMIER**. They are developing a potential new treatment for subjects living with CMT1A. The purpose of the **PREMIER** study is to find out if a study drug, called PXT3003, is safe and effective at treating people with CMT1A. It is hoped that PXT3003 can improve how the nerves function, thereby improving the symptoms of CMT1A.

Pharnext are currently looking for people age 16-65 years with a confirmed genetic diagnosis, who may be interested in joining the **PREMIER** study. The **PREMIER** study will take approximately 17 months to complete. During the study, PXT3003 will be compared to a placebo (an inactive or "dummy" drug). Subjects will be assigned to one of two treatment groups as follows:

1. PXT3003 taken orally, twice daily for 15 months
2. Placebo taken orally, twice daily for 15 months

The treatment groups will be selected at random by a computer. Subjects will be split evenly across the two groups, so they will have a 1 in 2 chance of receiving the study drug. The study is double-blinded, which means that for the entire study neither the subjects nor the study doctors will know if they are receiving PXT3003 or placebo.

Eligible subjects will attend the clinic every 3 months (6 visits in total), and the study team will contact them by telephone at least twice between these visits. They will also attend a final clinic visit, known as a Safety Follow-Up Visit, to assess their ongoing health and well-being.

For more information, visit www.premiercmt1a.com/ where you'll be prompted to answer **six questions** to check if you may be eligible for the **PREMIER** study. Based on your answers, a pop up will let you know if you are eligible and you can view a map listing sites that are enrolling patients with the contact information. **Or you can fill out the form** on this page and someone at **HNF will reach out to you or direct your email to** info@hnf-cure.org.

Brad Floyd

Brad Floyd – Team CMT Member, **Dad of Addie**

This is 50 and this is me in pain.

On Sunday, November 7, 2021 I was honored to represent Team CMT at the TCS New York City Marathon.

I signed up late with a short training window. When my Dad passed away I just couldn't lace 'em up and pretty much stopped training.

I thought of Addie and people with CMT and their daily struggles to do the most common tasks while facing ridicule for being slow and having challenges most can't understand.

I picked up David Goggins book and read a page I had highlighted. I'll paraphrase: "when you persist past the point when pain fully saturates your mind, boundaries dissolve. . ."

I have always said I have a different relationship with pain than most as do many people with hidden illnesses whose struggle goes unnoticed.

If they can persevere, so can I.

In the race, as expected, the pain came hard and early. "I felt as much gratitude as pain . . .as much appreciation as discomfort." Doing this race is an honor.

This is the largest marathon in the world.

It is special. The crowds were just amazing with street jam packed from the first mile to the end.

I took in the excitement, the energy, and absorbed the struggle for more than 4 hours. I crossed the finish and collapsed.

My brother and sister were there to pick me up for which I am grateful.

I want to thank all of you who donated and made this possible as well as Hereditary Neuropathy Foundation for this opportunity. I want to thank Karen Lanciault for being the CMT Ambassador and best Mom to Addie.

I want to thank the Hereditary Neuropathy Foundation for this opportunity.

Push past boundaries. Show compassion for others. Be a better version of yourself.

Thank you Brad for you dedication to Team CMT

CLICK HERE TO JOIN TEAM CMT!

Congrats to Team CMT Member Kat Guenther for completing two half marathons!

Kat Guenther

Team CMT Member

Hill Country Trivium November 7, 2021 which they proudly boast to be the hardest Marathon in the hill country & the Rock 'n' Roll Running series in San Antonio December 5, 2021.

TD Bank Five Boro Bike Tour

HNF is excited to announce that we have been selected for the 12th year as a charity partner. The tour will be May 1, 2022 in New York City.
TO PARTICIPATE OR IF YOU HAVE QUESTIONS CONTACT: courtney@hnf-cure.org.

Join Team CMT!

If you, a family member, or a friend would like to help support HNF's programs and fund vital research, please consider joining the team. **TO LEARN MORE CLICK HERE!**

Top 3 Webinars!

Since the inception of CMT-Connect video series here are the top three webinars:

1. **Cannabis & CBD for CMT:** CLICK HERE!
2. **Surgery & CMT with Dr. Glenn Pfeffer:** CLICK HERE!
3. **Pain Series Pt. 1 with Dr. Wayne Berberian – Do I Need Surgery?:** CLICK HERE!

Have a topic you would like CMT-Connect to cover? **EMAIL:** CMTCONNECT@HNF-CURE.ORG

PAST WEBINARS

- Guided Meditation
- Work From Home Job Training & Placement
- CMT & Telemedicine
- Align with Happiness
- CMT & Capture Proof
- CMT & Genetic Testing
- CMT & Covid-19
- Healing from the Inside Out
- CMT Resources with Inspire
- Dating & CMT
- How to Exercise in the Pool with Bernadette Scarduzio
- accessibleGO.com: A New Way to Travel with Disabilities
- Bemer Technology
- Panetta Physical Therapy
- CMT & Balance
- CMT & Your Nutrition
- CMT&Me App
- CMT & Exercise
- Mobi Mats
- Surgery & CMT
- CMT & Finances
- Ability360 Sports & Fitness Center
- Active Hands
- Jamal Hill ~ Paralympic Swimmer
- CMT & Microcirculation
- Cannabis & CBD for CMT
- CMT & Canine Companion
- Mental Health & CMT
- Pain Series: Part 1 - Do I need Surgery?
- Pain Series: Part 2 - Nutrition
- Accessible College
- Family Planning #1: IVF
- AFOs for CMT
- SORD Deficiency

VIEW PAST WEBINARS:
Click Here!

WTF is CMT?

Order your *NEW* CMT Info Cards: $25 Donation (24 pack)

Have you ever been asked annoying questions like:

- What's wrong with your legs?
- Do you really need that scooter?
- Why are you parking in that handicap spot?
- Are you drunk?

Well, have we got the answer for you!

Introducing, "WTF is CMT" INFO CARDS!

Simply share any of our 6 assorted cards and TaDa! They'll be magically transported (via QR code) to our WTF is CMT page.

So next time you're contemplating running someone over with your scooter or wheelchair, stop… and hand them a card instead and say WTF!

CLICK HERE TO VIEW VIDEO AND ORDER CARDS ➜

Ask Lainie

Dear Lainie,

I have CMT and just got divorced. I'm worried that I will never find someone now that I wear AFOs and my disabilities are more apparent than they were when I was young and single. Do you have any dating tips?

Thanks for your help!

– Lisa P. from Philadelphia

Dear Lisa,

I can absolutely relate and help! When I got married in 1997, I thought, as most new brides do, that it would last forever. I had known my former husband since college. The fact that I had a progressive neuromuscular disorder was never a factor in our relationship nor in its eventual demise. He was supportive when I got my first pair of leg braces. He was patient and helpful when I needed his arm for balance. He was (and still is) a great guy.

Ten years, two kids, and several wrinkles later, I re-entered the dating world in my 40's. It was exciting, scary, disappointing, depressing, and even comical at times. You can read about some of my experiences and tips **in this Trend-Able Post.**

I blamed CMT for my inability to find love again and used it as a defense mechanism for coping with the "ghosting" and rejection that inevitably happens (to many daters today irregardless of ability) when we put ourselves out there. It wasn't until I learned to accept my perfectly imperfect self and owned the numerous positive attributes I'd bring to a relationship, that I eventually found and married the man of my dreams.

In episode #5 of the **EmBRACE It with Lainie & Estela Podcast**, we discuss dating & the red flags we both learned to stay clear from when dating with a disability.

For more dating tips, you can also read **this fabulous guest post** by Rebecca. a young and single LA woman with CMT.

Xo

Lainie

Lainie Ishbia, MSW
Disability Speaker & Blogger
Trend-ABLE

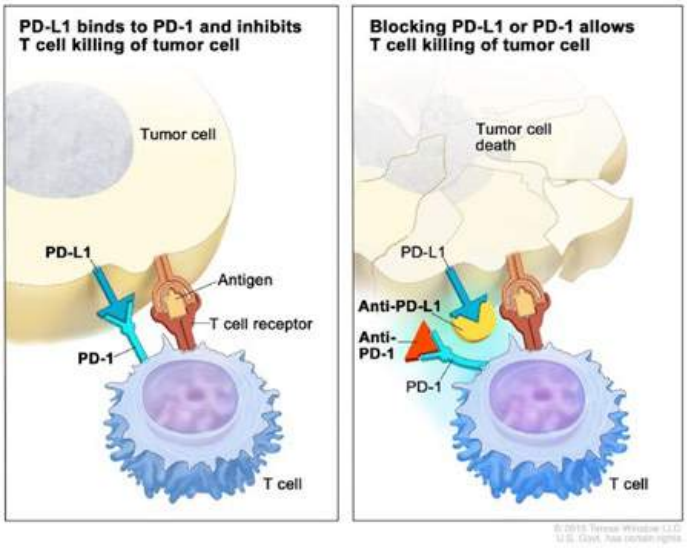
NEUROTOXIC DRUG CAUTION

CORINNE WEINSTEIN, CLINICAL ONCOLOGY PHARMACIST AT CANCER CENTERS OF COLORADO-GOOD SAMARITAN, DENVER, COLORADO

Immune Checkpoint Inhibitors (ICIs)

Immune Checkpoint Inhibitors are a type of immunotherapy used to treat a variety of cancers. Immunotherapy is a modern approach to cancer treatment that harnesses the power of your own immune system to kill cancer cells. Your immune system's main job is to distinguish normal, healthy cells from abnormal cancer cells. Proteins on the surface of cancer cells (PD-L1 for example) interact with proteins on immune cells (PD-1 for example), specifically T cells. These interactions are called immune checkpoints. Cancer cells prevent attack by "blindfolding" the immune cells. ICIs remove this blindfold, allowing your immune system to attack cancer cells (See Image Below).

"Boosting" the immune system with immunotherapy, however, can cause your immune cells to attack healthy cells in the body as well, including nerve cells. Varying types of neurological complications can occur with ICIs, affecting both the central and peripheral nervous system. Peripheral neuropathies (which is most relevant to CMT patients), however, occur in less than 1% of patients and vary in severity and type. They include demyelinating, axonal sensory motor, and pure sensory neuropathy. They may have an acute onset similar to Guillain-Barré syndrome (GBS) or be slow to develop. The risk is similar among ICIs, with the highest risk occurring when nivolumab (Opdivo) and ipilimumab (Yervoy) are used together. Therefore, although rare, patients with an underlying hereditary neuropathy should use caution when utilizing these agents. If symptoms do develop or worsen, the ICI should be discontinued and steroids and/or immunoglobulin may be given to mitigate symptoms. As always, the risk/benefit of ICIs should be discussed with your oncologist.



PD-1/PD-L1 Inhibitors
Nivolumab (Opdivo)
Pembrolizumab (Keytuda)

CTLA-4 Inhibitors
Ipilimumab (Yervoy)

Highest Risk
Ipilimumab (Yervoy) + Nivolumab (Opdivo)

Immunomodulatory Drugs (IMiDs)

Immunomodulating drugs are medications that work directly on the immune system by either turning up or turning down certain proteins. These drugs also exhibit antiangiogensis effects, meaning they work to cut off blood supply to tumors, limiting its ability to grow. They are used to treat a variety of cancers including multiple myeloma and certain types of lymphoma.

While all three of these medications pose a risk of peripheral neuropathy (PN), thalidomide has been shown to pose the highest risk. One study found the incidence of severe grade peripheral neuropathy was 6% in patients receiving single agent thalidomide, while peripheral neuropathy of any grade was reported in 28% of patients. The risk is closely related to the cumulative dose and the duration of treatment, with peripheral neuropathy occurring in up to 70% of patients treated with thalidomide for more than 12 months. PN can also occur from relatively short-term therapy and may be irreversible. Overall risk increases for patients taking multiple neurotoxic medications, elderly patients, and those with preexisting neuropathy, such as CMT. The exact mechanism by which these medications cause neuropathy isn't completely understood, but one study suggested that thalidomide may have direct effects on the dorsal root ganglia (DRG, a group of cell bodies responsible for the transmission of sensory messages), leading to its degeneration.

(Continued next page)

NEUROTOXIC DRUG CAUTION

In general, lenalidomide and pomalidomide are considered much less neurotoxic, with a significantly lower risk of peripheral neuropathy compared to thalidomide. However, a risk of peripheral neuropathy still exists. One study, examining patients receiving lenalidomide and dexamethasone, demonstrated severe grade neuropathy developed in 1.7% (3 of 177) of patients. Given this and the overall more favorable safety profile of lenalidomide and pomalidomide, these agents are more widely used compared to thalidomide.

Patients with an underlying hereditary neuropathy should use extreme caution if these medications are used. Again, a discussion of the risks and benefits of therapy should always be discussed with your hematologist or oncologist. If alternative therapies are not an option, limiting the dose and duration of therapy, whenever possible, may help reduce the risk of neurotoxicity.

IMMUNOMODULATORY DRUGS (IMIDS)

1. Lenalidomide (Revlimid)
2. Pomalidomide (Pomolyst)
3. Thalidomide (Thalomid)

TAKE AWAY POINTS FOR CMT PATIENTS

1. Avoid use of IMiDs whenever possible. If the benefit of therapy outweighs the risk, lenalidomide and pomalidomide have a lower risk of neurotoxicity.
2. Limit the dose and duration of therapy whenever possible.
3. Always disclose your CMT to your oncologist/hematologist. Alternative therapies may be available with a lower risk of neuropathy.

ASK THE EXPERT

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: I was diagnosed with CMT in December 2019. I would like to know what you would recommend to your patients regarding the COVID-19 vaccination. Do you recommend getting vaccinated and if so, which brand do you recommend?

A: Dr. Kafaie: While considering a medication, we evaluate its risks versus benefits. COVID-19 infection, unfortunately, has severe morbidity and mortality, and the benefit of the COVID vaccine in lowering the risk of transmission is well established. There is not data available to comment if it has any added side effects in patients with CMT compared to the general population. With the lack of evidence for its potential extra side effects in CMT and its established benefits, I think it is reasonable to recommend vaccination in CMT patients. From this perspective, all available brands should be okay. I hope this helps.

HAVE A QUESTION?
www.hnf-cure.org/ask-the-expert

New Center of Excellence: University of Toronto

HERNAN D. GONORAZKY, M.D. CSCN DIPLOMATE (EMG); ASSISTANT PROFESSOR, FACULTY OF MEDICINE); THE HOSPITAL FOR SICK CHILDREN | DIVISION OF NEUROLOGY, DIRECTOR, NEUROMUSCULAR FELLOWSHIP PROGRAM, UNIVERSITY OF TORONTO

Dr. Hernan Gonorazky has been a staff neurologist at the Hospital for Sick Children as the neuromuscular program director since 2019.

He received his MD from the University of Buenos Aires in 2007. As an undergraduate, he worked as a volunteer at the Argentinean Foundation for Myasthenia Gravis where he was involved in the care and treatment of patients, as well as clinical research.

He did his fellowship in adult neuromuscular disorders at the Italian Hospital of Buenos Aires where he had the opportunity to be engaged in the care of adults. In 2014, he became a clinical and research fellow at the Hospital for Sick Children.

Since joining the team at SickKids as an academic-clinician in 2014, his focus has been on finding new diagnostic methods in congenital myopathies, as well as improving care and education of neuromuscular disorders.

He is currently the co-director of the Spinal Muscular Atrophy program at Sickkids and has dedicated much of his effort to spreading education about neuromuscular disorders.

Q: WHY IS CMT YOUR PASSION?

CMT is the most common group of neuropathies in the pediatric population. Although some of the causing genes are quite common, other ones are rare genes without much information on natural history and outcomes. As a physician caring for children and youth with these conditions, I feel a strong responsibility to not only provide care, but better understand about the complexities behind their condition. If we do not lead these efforts, then who will?

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

At SickKids, we work each day to provide the best in child and family-centered care. For patients with CMT, our multidisciplinary clinic will follow them for 6 to 12 months depending on the complexity and severity of the condition. This clinic consists of a physician, nurse practitioner, EMG/NCVs technician and orthopedic specialist. In more severe cases, we will coordinate care with the respiratory and orthopedic clinics. All of the patients will also be connected with physical therapy or occupational therapy in their community or within a rehabilitation center.

Q: WHAT DO YOU LOVE MOST ABOUT YOUR PRACTICE?

Understanding the complexity and natural course of CMT to help care for patients with this condition is of utmost importance to our team. I also enjoy collaborating with my colleagues in the translational research laboratory to study and develop better strategies to treat this group of disorders.

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

A primary health-care provider must provide a referral to the Neuromuscular Clinic at SickKids. Once that has been obtained an appointment can be arranged.

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 living with CMT and families with inherited neuropathies.

ARIZONA

Barrow Neurological Institute
602-406-8144
Dr. Shafeeq S. Ladha

ARKANSAS

Arkansas Children's Hospital
*Pediatric Center of Excellence
501-364-1850
Dr. Aravindhan Veerapandiyan

CALIFORNIA

Cedars-Sinai Medical Center
310-423-4268
Dr. Richard Lewis

Stanford Neuroscience Health Center Neuromuscular Clinic
*Pediatric Center of Excellence
650-723-6469
Dr. Robert Fisher (Adults) Dr. Joy Day (Pediatrics)

UCLA Department of Neurology
310-794-1195
Dr. Halabi Anasheh

COLORADO

Children's Hospital Colorado
*Pediatric Center of Excellence
720-777-2806
Dr. Michele Yang

CONNECTICUT

Hospital for Special Care
Charles H. Kaman Neuromuscular Center
860-612-6305
Dr. Kevin Felice

FLORIDA

University of Florida (UF) Health
352-294-5000
Dr. James Wymer

University of Miami
Professional Arts Center (PAC)
305-243-7400
Dr. Mario Saporta

AdventHealth Neurology at Winter Park
407-303-6729
Dr. Nivedita Jerath MD, MS

ILLINOIS

Ann and Robert H. Lurie Children's Hospital of Chicago
*Pediatric Center of Excellence
312-227-4471
Dr. Vamshi Rao

Rush University
312-942-5936
Dr. Ryan Jacobson

UI Health Hospital and Health
Sciences System
Outpatient Care Center
312-996-4780
Dr. Charles Abrams

KANSAS

University of Kansas Medical Center
Landon Center on Aging
913-945-9926
Dr. Jeffrey Statland

MASSACHUSETTS

Brigham and Women's Hospital
Department of Neurology
617-525-6763
Dr. Anthony A. Amato

MICHIGAN

University of Michigan
734-763-2554
Dr. Dustin Nowacek

MISSOURI

St. Louis University Medical Center
314-977-6177
Dr. Jafar Kafaie

MU Health Care
612-624-7745
Dr. Karim Salame and Dr. Vovant Jones

MINNESOTA

M Health Fairview Clinics & Surgery Center – Maple Grove
612-624-7745
Dr. David Walk

NEW JERSEY

Goryeb Children's Hospital
*Pediatric Center of Excellence
973-971-5700
Dr. Jahannaz Dastgir

Hackensack University Medical Center
Neuroscience Institute
551-996-8100
Dr. Florian Thomas

NORTH CAROLINA

University of North Carolina
984-974-4401
Dr. Rebecca Traub

Atrium Health Neurosciences Institute-Charlotte
704-446-1900
Dr. Urvi Desai

NEW YORK

Columbia University
Department of Neurology
212-305-0405
Dr. Thomas Brannigan

TEXAS

Austin Neuromuscular Center
512-920-0140
Dr. Yessar Hussain

OREGON

Oregon Neurology
541-868-9430
Dr. Michael Balm

WASHINGTON

St. Luke's Rehabilitation Institute
509-939-8079
Dr. Gregory Carter

University of Washington Medical Center
206-598-0058
Dr. Nassim Rad

Please visit our Center of Excellence for more information.
www.hnf-cure.org/centersofexcellence

*Pediatric Center of Excellence

HEREDITARY NEUROPATHY FOUNDATION

1641 3rd Avenue
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hnf-cure.org

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

www.hnf-cure.org

HNF
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Events

Get Involved and Join us at an Upcoming HNF Event

THRIVE Patient-Centered Charcot-Marie-Tooth Summit
March 5-6, 2022

TD Bank Five Boro Bike Tour
May 1, 2022

Movement is Medicine™ Summit
November 4 -5, 2022

TCS New York City Marathon
November 6, 2022

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