HEREDITARY NEUROPATHY FOUNDATION

FALL/WINTER 2019

cmtupdate

A Successful Movement is Medicine™ Summit

Pg. 2

TRIAD
Thinking Outside The Box
Pg 8

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

www.hnf-cure.org

CMT UPDATE
FALL/WINTER 2019

Allison T. Moore
Founder/CEO, Hereditary Neuropathy Foundation

Dear Friends,

As we look back on 2019, we are humbled by all the support we have received from our community and are proud of the progress we made again this year. Our innovative patient-centered research program continues to break new ground by implementing research efforts for therapies that will help our patient community today.

We hear you and we take action!

Whether it is at our annual summits, via our online Inspire CMT Community or from the data we collect in our patient registry, Global Registry for Inherited Neuropathies (GRIN), our members with CMT share their daily struggles with pain, weakness, depression, or difficulty exercising due to fatigue. As a severely affected patient myself, I deal with these issues on a daily basis and know all too well the toll they can take on your physical and mental health.

As the Founder and CEO of HNF, though, I just couldn't accept that this should be a CMT patient's reality. Knowing that *anything* is possible when you put your mind to it, we mobilized our HNF team along with some passionate volunteers and created our newest program: Movement is Medicine™. On November 7 and 8, HNF brought together leading CMT experts in Physical Therapy, Occupational Therapy, Kinesiology, and physical fitness to engage our participants in a groundbreaking full two day event.

We are grateful for all the individuals who were able to attend. We are eagerly planning our 2020 Movement is Medicine™ Summit and hope you will be able to join us! Make sure you save the date: November 13 and 14, 2020.

As many of our programs often incorporate research, we conducted, in collaboration with Arizona State University and ProKinetics, a pilot gait study called "**Digital Assessments of Balance and Gait in people with Charcot-Marie-Tooth Disease**". This study captured critical patient mobility, balance and gait data that we hope will improve clinical trial design for upcoming clinical trials. We thank all that participated in this groundbreaking study, and look forward to sharing the results with you as soon as they are published!

HNF's research and industry collaborators recognize that exercise is an adjunct therapy to support disease modifying drugs and cures for CMT. HNF has taken the findings and feedback from the summit and will continue to build a research driven exercise model with the goal of bringing exercise therapies to you that can help maintain or even increase function.

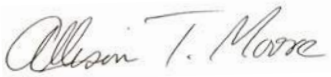
In this issue, please make sure to read up on the exciting Therapeutic Research in Accelerated Discovery (TRIAD) research projects that continue to expand to other types of CMT. We have been working with some of the top gene therapy experts in the world since 2017, and are energized by the new scientific collaborations we have made to advance our gene therapy program. HNF co-founded GRIN back in 2013 with Hannah's Hope Fund and continues to work alongside Co-Founder Lori Sames to advance gene therapies for multiple types of CMT. Lori Sames is a gene therapy pioneer that, from her kitchen table, successfully advanced a gene therapy to save her daughter Hannah's life and hundreds more that have a rare type of Inherited Neuropathy called Giant Axonal Neuropathy.

In Lori's words, "We can now accelerate the process exponentially. With good science, an aggressive timeline and funds, new gene therapies can be in humans in 3 years!".

So let's get moving!

If you would consider making a year-end contribution, I would really appreciate it. Every dollar matters and helps HNF to better serve you and your families.

I wish you and your family a happy and healthy New Year!



A SUCCESSFUL **MOVEMENT IS MEDICINE** SUMMIT

The Hereditary Neuropathy Foundation would like to thank all the sponsors who made the Movement is Medicine™ Summit possible. Pharnext, Acceleron Pharma, Vector Sports by Kayezen, Athena Diagnostics, BioTek reMEDys, ProtoKinetics, Vantage Mobility International (VMI), Lacuna Botanicals, Ortho Rehab Designs: Helios Bracing, Fior & Gentz, Active Hands, Ottobock, Champlain Valley Dispensary, and Bombas all graciously provided financial support and products for the Movement is Medicine™ Summit.

HNF is grateful for all the individuals who helped to make this incredible Summit possible and who are making a difference in the lives of our courageous attendees.

The HNF team is already planning next year's Movement is Medicine™ Summit for November 2020.

We are grateful to all the patients who participated in this groundbreaking event! The wonderful feedback we have received makes all of the hard work and planning well worth it.

HERE IS WHAT SOME OF OUR ATTENDEES HAD TO SAY:

"Watching my wife get involved with activities, seeing her spirit open up by just being around others with her disability made this event our best of the year! And, we got to meet our CMT heroes!!!"

– **Kenneth**

"It was such an incredible weekend attending the Movement is Medicine™ Summit. I feel more alive and inspired. I loved being with other people who have CMT and are working on their health and well being. It was such a positive, fun, and safe space. I loved the Ability 360 location, learning about different exercises I haven't done before, and meeting wonderful people. I fell in love with the HNF organization. I finally feel like I have found my tribe."

– **Cheryl**

"Empowering, Life-Changing, Life-Saving and Good for the Soul!"

– **Jennifer**

"This summit gave my daughter and me such inspiration and hope. We learned much more about CMT and how to combat its progression. We have put everything we learned into action in our everyday lives now. Thank you."

– **Terry**

"The Summit was a valuable experience just by bringing together a wide array of individuals with a shared lifestyle based on our disease. The workshops taught exercises that I can do on my own to help me stay mobile."

– **Janet**

"Patients with CMT need information, and there are not enough physicians who understand the disease. The Movement Is Medicine™ was a tremendous resource."

– **Bill**

"I really felt comfortable in my body being around people with similar struggles (balance, gait, strength and other ailments caused by my disease). The fitness instructors were sympathetic to my limitations, which I really appreciated."

– **Laurie**

"I feel less lonely. I feel braver and more accepting of myself. I saw and met people with some of my visible issues and have more perspective about how other people react to me. I feel less self-conscious about returning to my own life."

– **Rojean**

"I learned that I can exercise in many different ways without over-exerting myself and causing pain. I was able to feel like myself and didn't feel like I had to act."

– **Taylor**

"The Summit exceeded my expectations—from meeting and learning about other CMT warriors who were willing to share their experience, to attending all the educational sessions, which provided such valuable information—I am going home with new friends and hope to continue my CMT journey."

– **Amanda**

"Like meeting family you never knew you had, but always wanted"

– **Jeff**

PATIENT'S VIEW OF HNF **MOVEMENT IS MEDICINE** SUMMIT

CHERYL SHERMAN, CMT 1B PATIENT

Shortly after the Summit, we received a letter from Cheryl Sherman, who attended our Movement Is Medicine™ Summit. Cheryl has CMT 1B and was diagnosed 23 years ago when she was 20 years old. She lives in Sacramento, CA with her husband and 2 cats.

Here is her feedback:

I had the absolute honor and pleasure of attending HNF's Movement is Medicine™ Summit on November 8-9, 2019. This was the first time I attended a conference on CMT where I felt inspired and energized with the breakout sessions I attended. I also felt totally connected with the individuals I met who either have CMT or are supporting a loved one who has CMT.

I absolutely loved where the summit was being held: Ability 360 in Phoenix, AZ. This state-of-the-art workout facility was designed for people with disabilities! Everywhere I went was accessible! Even the rooftop deck was accessible, which is where we had a cocktail party on our first night. The facility includes a lap pool, warm water therapy pool, hot tub, and accessible locker rooms. It had a large sports court for a variety of activities including wheelchair rugby, adaptive basketball, adaptive dance classes, etc. There is also an upstairs area with workout equipment and free weights for cardio and strength training. Plus, rooms for other types of classes like yoga. And to top it all off, they had a rock climbing wall that was adaptive!

I loved the way the summit was put together. There were breakout sessions each hour where you could participate in many different topics on improving your health and well being. There were yummy meals provided (which I so appreciated since I was ravenous after the intense workout in the morning) and then more breakout sessions in the afternoon. There were also vendors you could talk to, such as AFO companies like Ortho Rehab Designs, which makes the Helios braces, as well as Fior & Gentz and Ottobock and the fabulous Lainie Ishbia from Trend-Able, selling cute shirts. I thoroughly enjoyed socializing with other people who have CMT or who have loved ones who have CMT.

My husband and I attended on the first day Aqua Yoga, Core Tabata, The ABC's of AFO's, Nutrition for the Nervous System, and Aqua PT taught by Bernadette Scarduzio. We also really enjoyed hearing Jamal Hill speak to us about his story and journey to compete in the 2020 Paralympics!! On the second day, I participated in a series of exercises where researchers were collecting data on Balance and Gait exercises from people who have CMT. Then we attended breakout sessions for the intro class for Made for Motus by Christopher Dito, had a one-on-one demonstration of how to use the Kayezen Vector system, Crowd-Design Workshop for Clinical Trials, Yoga for All, and Adaptive Dance.

I felt the breakout sessions I attended were informative, fun, and inspiring. I enjoyed learning how to do tree pose in the water (which I can not do on land) during the Aqua Yoga class. I had a lot of fun doing Core Tabata, which is circuit training with 7 stations including abs, planks, and a couple of different arm exercises with weights and balance exercises. I have done most of these exercises to some degree or another, but it was fun to put them all together in a circuit, where you did one station for 20 seconds with a 10-second break and then did that for 4 sets before moving onto the next station.

The Aqua PT with Bernadette Scarduzio was a lot of fun. First of all, I thoroughly enjoyed getting to meet my hero, and then it was also great getting to learn different ways of working out in the pool, including moving the dumbbells around at a faster speed than I am used to, which really gets the heart rate up and gives a nice burn as well.

I really enjoyed the Yoga for All, where there were 4 instructors in the room helping people as needed. There were plenty of supplies available like bolsters and yoga blocks. But I really enjoyed having the instructors help me out when I couldn't do some of the exercises, such as rotating my feet around. It was very sweet and helpful to have one of the instructors come over to help rotate my feet for me, and then he also helped me keep my balance when I was trying to do the warrior pose.

But I have to say my favorite breakout session was the Adaptive Dance class (i.e. a Zumba class). It was lead by Justin, an amazing instructor who works at Ability 360. Justin brought an attitude of fun, playfulness and high energy to his class. He had these gold pom pom's that he used to gesture while calling out dance moves. His high energy, fun and playful attitude, and his encouraging spirit, plus those awesome gold pom pom's, gave me the space to dance freely. I have loved dancing ever since I was a little girl. However after my CMT started to affect my ability to dance fluidly, I became more self-conscious when dancing. I am not as coordinated as I used to be and my body moves slower than my mind knows how the dance moves should go. But I have to say that I felt truly free and self expressed dancing with this amazing group of people. It made such a difference to me to be with people who also had limitations but were still shaking it on the dance floor. And, of course, having an instructor like Justin who made it so inviting made the world of difference too. It really felt so good to have the space of no judgment while dancing.

My experience the entire two days reinforced that there was the space of no judgment in all of the breakout sessions I attended!! It is really important to have a safe space to try out new exercises without judgment. The Movement is Medicine™ Summit 100% accomplished that in my mind.

To me, the best part of the two days was meeting so many incredible people, including people who are my heroes in the CMT Community. It was great to be with so many like-minded individuals who are positive and taking on their health and well being to the best of their ability based on the limitations we all face. I loved meeting every single person, and can not wait for next year!! It is the first time where I can say I have found my tribe!!

I do wish I could have attended all of the other breakout sessions as well. There were so many interesting workout sessions to attend. The input I got from other participants on these other classes showed me that everyone had a really good time in whatever session they were in. There was just not enough time to attend every single topic available. And there were some breakout sessions where at the time I did not have an interest in and now I do. For instance, I had seen on the schedule that you could do rock climbing. That wasn't even in my realm of possibility due to my limiting thoughts around being able to actually rock climb. But after seeing pictures of people that I met at the conference that took on rock climbing, I can now see that even I could participate in that! So, next year I look forward to trying adaptive rock climbing!

As you can see, I really did have a blast at the Movement is Medicine™ Summit. It was truly life-changing in so many different ways. I will absolutely be attending next year. I hope you will join me. HNF's Movement is Medicine™ Summit is an event not to miss!

TRIAD
THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY

HNF tackles the **hidden mysteries behind the genetic diagnosis** of CMT mitochondrial diseases

C12orf65 gene, a rare form of CMT also known as Leigh Syndrome is robbing young children of a normal life even beyond the classic CMT symptoms, including autonomic distress, blindness and in some cases death. Jaxson Flynt, was diagnosed with C12orf65 on October 14, 2014, a terminal form with progressive damage to the central nervous system. It is caused by a defect in the function of mitochondria within the cells of the body. Jaxson is a happy boy who loves to play basketball and spend time with his mom (Lindsey), dad (Garret), sister (Madison, 16) and two labradoodles (Remy and Reese).

Jaxson is non-verbal, has limited vision and fine motor skills and receives his nutrition through a g-button. However, Jaxson has met many milestones the doctors thought would be impossible. They thought he would never walk, yet he can get around with the best of them.

Lindsey reached out to HNF Scientific Advisory Board Member and gene therapy expert, Dr. Steven Gray of UT Southwestern looking for answers and was hopeful that gene therapy will cure their little boy.

Fortunately, HNF has been working with Dr. Steven Gray since 2017 and he had already begun work on a gene therapy approach for C12orf65. So it was only natural for the HNF to embrace the Flynt's request by having them join us in raising the funds needed to cure Jaxson and others.

"*Our goal is to slow the progression of this disease while working towards a cure for Jaxson and future generations. Gene therapy looks to be a promising life extending treatment for Leigh Syndrome (C12orf65). The field of gene therapy has gone through remarkable breakthroughs these last few years and offers incredible promise for rare genetic diseases such as C12orf65.*"

— Lindsey Flynt, Jaxson's Mom

TO DONATE:
www.hnf-cure.org/jaxsons-cure

Like with many of these rare diseases, it's not an easy task with many challenges to overcome, but we are hopeful. To date, HNF in collaboration with Dr. Robert Burgess at Jackson Labs, worked on the development of a mouse model of C12orf65, but once the mouse developed C12orf65, the pups would die. This was super tricky so Dr. Burgess quickly re-evaluated the path and engineered a cell model as an assay to test the hypothesis that gene therapy could cure this disease.

The assay was developed based on the mutation C12orf65 of our board member Debi Houliares son, Zachary, who has CMT6. Dr. Robert Burgess was able to manipulate this model to replicate C12orf65 to establish proof of concept that gene therapy be a viable path for a cure.

Debi, an HNF board member since August 2014, has championed C12orf65. To date, Debi has raised the funds to support the development of a preliminary natural history study of C12orf65, developed the AAV virus vector at Dr. Steven Gray's laboratory (formally at UNC Chapel Hill). Stay-tuned as we announce the next steps in 2020.

TO DONATE:
www.hnf-cure.org/zachsteam

CMT&ME APP STUDY

Did you know you can now store your CMT data at your fingertips? The CMT&ME app, developed by Vitaccess, has updated the app to include a brand-new medical profile. To update the app, go to your app store and click update.

In collaboration with our industry partners Pharnext and Vitaccess, we are thrilled to bring to you new features of the CMT&Me App.

"*This two-year study, which has been launched in 6 countries, was built for you and by you, the patients*," says Dr. Youcef Boutalbi, Pharnext Medical Affairs Director at the groundbreaking HNF's Movement is Medicine™ Summit Crowd-Design Workshop on November 8-9, 2019.

Currently this is the only mobile App for patients to record details on their diagnosis, list current or prior medications or participation in clinical trials, and log in regularly to report on symptoms, such as pain, falls, sleep, and so much more.

This vital partnership gives our members the opportunity for their voices to be heard! By sharing your daily or weekly experiences and challenges, you are participating in very important research that will help Pharnext and the entire CMT community accelerate therapies to address your unmet medical needs, expand the knowledge of what you, as patients, face. Most importantly, it will provide a portal to store your data to refer back to or even share with your doctors.

Allison Moore, Founder and CEO of HNF, has worked closely with Dr. Mark Larkin, Founder and CEO of Vitaccess, to disseminate CMT&Me research findings at global medical conferences to increase awareness of the CMT patient experience and advise regulators and payers (ie. insurance companies) on the impact CMT has on quality of life. The latest poster sessions Mark and Allison were highlighted at the AANEM annual conference in Austin, TX.

- **Diversity in the Charcot-Marie-Tooth Disease Population in the United Kingdom and United States:** Insights from a Digital Real-World Observational Study
- **Treatment of Charcot-Marie-Tooth Disease in the United Kingdom and United States:** Insights from a Digital Real-World Observational Study

The CMT&Me App has shed additional insights into this grossly underserved community and the unmet need of medical care and treatments for our patient community. Pharnext, Vitaccess and HNF recognize the urgency of getting drugs approved and commercialized for CMT, so don't wait, join today!

CLICK HERE FOR MORE INFORMATION:
www.hnf-cure.org/living-with-cmt/cmtme-study

DOWNLOAD THE CMT ME APP TODAY!
www.hnf-cure.org/cmtme-study

TRIAD
THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY

THINKING OUTSIDE THE BOX

A long road to discovery of a potential novel treatment for a rare form of Charcot-Marie-Tooth (CMT) neuropathy

FLORIAN P THOMAS, MD, PHD, DIRECTOR, HEREDITARY NEUROPATHY CENTER OF EXCELLENCE; CHAIR, DEPARTMENT OF NEUROLOGY & NEUROSCIENCE INSTITUTE; HACKENSACK UNIVERSITY MEDICAL CENTER & HACKENSACK MERIDIAN SCHOOL OF MEDICINE

When in 1998 I met a large, five-generation family with hereditary neuropathy, little did I know that this would lead to a research effort now entering its third decade. A pattern emerged among the over 15 affected relatives: While their clinical presentation was one of typical CMT, their electrical studies did not fit the neat distinction of demyelinating CMT1 with slow conduction speeds and axonal CMT2 with near normal speeds, but were smack in the middle, i.e. intermediate. At the time, only a handful of genetic tests were commercially available and they came back normal. So did this family have a novel subtype of CMT?

FIGURE 1:
Members of the US family with DI-CMT C and varying degrees of calf atrophy (with permission)

Around that time, at a scientific neuropathy meeting, I met the leaders of a renowned genetics group at the University of Antwerp, Belgium. The prospect of a novel CMT subtype piqued their interest and they offered to work with me identify the genetics in this family. It so happened that this group had become aware of another large family, this one from Bulgaria, whose member resembled the US family in also having intermediate conduction velocities.

Fast forward three years: In 2003 the American Journal of Human Genetics published our finding of a region on chromosome 1 which seemed to harbor a gene (as yet unknown) that was linked to having CMT in both families. Fast forward another three years: In 2006, Nature Genetics reported our discovery of the responsible mutations in a gene called YARS in humans & TyrRS in rodents. In 2016 and 2019, we reported a description of the clinical subtype, dubbed dominant intermediated CMT type C (DI-CMT C).

It was a surprise. The gene encodes a "housekeeping protein" called tRNA synthetase, which every cell uses for the basic function of linking the building blocks of proteins. How could a mutation in such gene only affect nerves? I had looked for problems in other organ systems than nerves in my patients, but found none. Our findings suggested that this protein played a role in axonal growth, which seemed to makes sense for a neuropathy. But how? In general, gene mutations cause disease through one or more than mechanisms: If the mutated protein no longer does its job, that can damage cells and organs. Or, a mutated protein can assume a new different function which can poison cells. We looked at two additional regions in YARS that are separate from the housekeeping function, one of which resembles a protein involved in inflammation, and another which promotes growth of blood vessels. It turns out we were barking up the wrong tree: There was no evidence that these parts of the protein were responsible for the neuropathy. This was frustrating, but by 2009 progress was made on a related project. When YARS was mutated in fruit flies (drosophila), it caused a neuropathy which prevented them from flying up when startled. So that made sense.

I met the leaders of a renowned genetics group at the University of Antwerp, Belgium. The prospect of a novel CMT subtype piqued their interest and they offered to work with me identify the genetics in this family.

Yet it took another 10 years till a collaboration between folks at University of Antwerp and Scripps Research in California hit pay dirt. In a study just published in Nature Communications we show that the YARS mutations in our families makes the protein toxic when cells are under stress. When cells are "happy" (so to speak), YARS protein is in what is called the cytoplasm, the compartment where cells. break down carbohydrates or lipids to produce energy, make proteins that contract in muscle cells or neurotransmitters in nerve cells. But when cells are under stress, YARS protein moves into the nucleus where it impacts which genes become activated. It turns out that the mutated YARS protein that is linked to neuropathy in both humans & fruit flies, functions differently from the normal version when inside the nucleus, and significantly changes how the cell works. And when in the fruit fly the mutated YARS is blocked from entering the nucleus, neuropathy does NOT develop. So have we cracked the case?

The answer is 'maybe'. It turns out that our colleagues at Scripps & in Antwerp found that a known agent, Embelin, in common use for centuries in the Indian medical tradition, prevents the YARS protein from entering the nucleus and rescues the development of neuropathy in the fruit fly. Embelin is extracted from Embelia berries which are remotely related to cranberries, persimmon, blueberries, brazil nuts and scarlet pimpernels.

This discovery is not only exciting for people with DI-CMT C. Mutations in several other tRNA synthetases are also linked to CMT. At this point we don't know if similar disease mechanisms are active in those subtypes. Time and a lot of work will tell. Hopefully this will take fewer than 10 years. And such for such efforts to be successful, we depend on the enthusiastic support of families with CMT. For that I am grateful.

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TRIAD
THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY

HNFs Pediatric Neurology Round Table at Child Neurology Society Conference

Charlotte, North Carolina

Charcot-Marie-Tooth (CMT) is a childhood disease. It actually starts in utero and many children are affected very early in life. For that reason, on October 23, 2019, the HNF team hosted a cocktail hour and dinner at the Child Neurology Society Meeting to discuss the unmet needs of the pediatric CMT population with a select group of expert pediatric neuromuscular neurologists and other stakeholders. HNF presented critical data from the Global Registry for Inherited Neuropathies (GRIN), the Inspire CMT online discussion community and important findings from the Externally-led Patient Focused Drug Development Meeting (PFDD) for the FDA.

Pediatric and young adult CMT and inherited neuropathy patients can have difficulty with social and emotional challenges, which can have lasting effects throughout their adult lives: social withdrawal, loss of friends, inability to keep up with peers in school and sports, public humiliation, bullying and fear of the future. New treatments should focus on the following unmet needs: ability to walk unaided, ability to balance/stand, improved hand function, respiratory weakness and other autonomic neuropathy effects, fatigue and pain.

Our shared vision is to collect perspectives on treatment options and to support pediatric drug development for participants under the age of 18. HNF with its team of pediatric experts have developed a stellar program to support the pediatric unmet medical need that is crucial in improved medical care guidelines, therapy development and for the support of clinical trials.

STAY-TUNED AS WE UNVEIL THIS VERY IMPORTANT INITIATIVE IN 2020!

Digital Assessments of Balance and Gait in people with Charcot-Marie-Tooth Disease Study

HNF broke new ground with a new and exciting clinical study to help advance the improvement of treatment options utilizing digital technology to support clinicians on enhanced treatment outcomes and to add digital technology to clinical trial protocols.

We are grateful and thanks to the patients at our annual **Movement is Medicine™ Summit** that made this study possible.

The goal of the investigators at Arizona State University (ASU) was to utilize new and exciting digital technology - ProtoKinetics Zeno Walkway Gait Analysis System - to better understand the impact of CMT in a way that is meaningful to our everyday lives. Additionally, the Global Registry in Inherited Neuropathies pre-survey study questions will give us additional insight into how gait and mobility relates to pain, life satisfaction and social isolation.

"Having a deeper understanding of patient mobility can be obtained by incorporating quantitative and objective patient-centric information into routine care. This type of information can hopefully lead to better clinical trials."

– Dr. Mark Gudesblatt
Neurologist and Clinical Research Leader in partnership with HNF
South Shore Neurological Associates

Preliminary Analysis of the data suggest that confidence in balance and foot strength are associated with 6 minute walk distances. We are still in the process of analyzing and getting other data that could be useful.

The study is open for one year. To qualify, please take a few minutes to join Digital Assessments of Balance and Gait in people with Charcot-Marie-Tooth Disease Pre-Study Survey.

Help researchers help you and change the way that therapies are evaluated.

TAKE SURVEY HERE:
www.hnf-cure.org/registry/balance-and-gait-pre-study-survey

"We are excited to be working with HNF to develop robust treatment protocols and analytical reports that cover a wide range of data-sets that will better define patients' functional performance results with granular detail, thereby enhancing patient care and incorporating innovation solutions for CMT clinical trials."

– Dr. Edward Ofori
Director of the Pathomechanics & Neuroimaging Laboratory
Arizona State University

TRIAD
THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY

"Chronic Pain Assessment: Patient Perspectives"

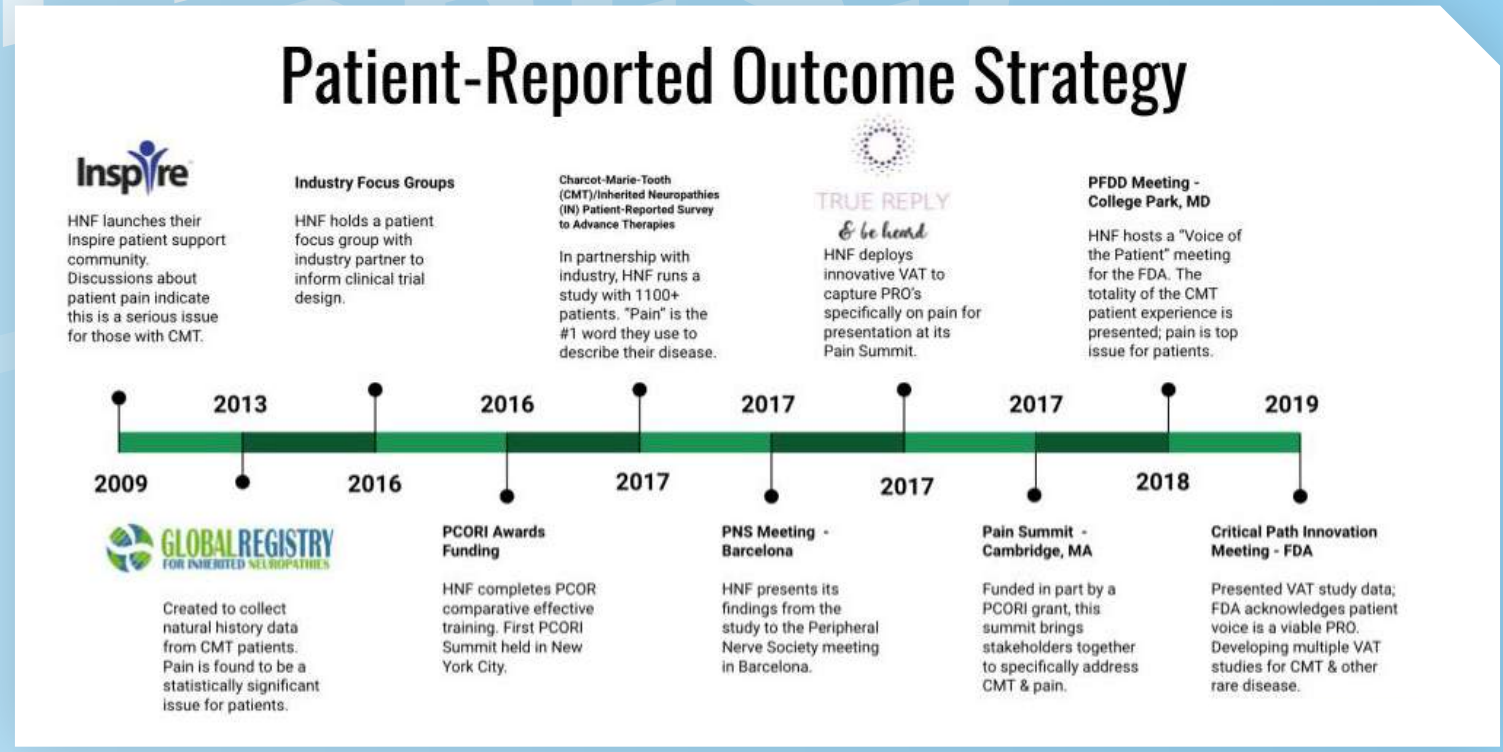
Allison Moore continues to participate in research initiatives to support CMT patients with chronic pain.

HNF is on the forefront of pain research to support CMT patients.

Our team is innovative and strategic, and most of our programs incorporate research efforts with a mission to identify gaps that hinder patient care and to move the needle in bringing treatments to patients.

Joy Aldrich is HNF's Advocacy Director and moderator of our online Inspire CMT community program (a safe haven for patients and families to communicate and share their experiences of living with CMT). She identified an overwhelming theme of pain amongst community members. She realized that patients and their families are in serious distress due to pain with little to no support from medical professionals or advocacy groups, and were often being told there is nothing to help them.

Allison Moore, HNF Founder/CEO, along with her team, took action and led the HNF groundbreaking CMT pain initiative to help the community.



HNF partnered with Acceleron Pharma and conducted a patient reported outcomes (PROs) research study to capture PROs to inform the Acceleron clinical team with data to design a Phase II, part 1, protocol to treat CMT1A, CMT1B and CMTX. This study has been instrumental to the CMT research community and has increased stakeholder engagement and funds to support critical programs for our patient community!

First, HNF quickly assembled a group of patients at Acceleron's headquarters in August, 2016 for a focus group to learn more about what matters most to patients when thinking about a treatment. From the initial data collected, they determined that more data was needed, and later launched a Patient-Reported Outcomes (PRO) Study through HNFs Global Registry for Inherited Neuropathies (GRIN). HNF launched the PRO study Charcot-Marie-Tooth (CMT)/Inherited Neuropathies (IN) Patient-reported Survey to Advance Therapies in February 2017. Not only did this study help Acceleron to design their pivotal Phase II, part 2, ACE-083 clinical trial, but it reiterated to us that pain is a huge gap that needs to be addressed for patient care and as an outcome measure in clinical trials.

This study has been groundbreaking. It not only informed Acceleron, but other pharma/biotech, industry, researchers, and the FDA on the impact that pain has on our patient community.

Taking additional action, Allison Moore, HNF Founder/CEO, joined the US Pain Collaborative, comprised of advocacy leaders from across the pain community: the Chronic Pain Research Alliance, For Grace, the International Pain Foundation, Reflex Sympathetic Dystrophy Syndrome Association, and U.S. Pain Foundation. With guidance from Voz Advisors, a biopharmaceutical consultancy, and with funding from Grünenthal Pharma, the collaborative wrapped up this year announcing the publication report, titled *Chronic Pain Patients Pinpoint the Need for Improved Methods to Assess Pain.* https://prn.to/2sAv7uf

The report is, in part, informed by a 2018 survey designed to understand the patient perspective on how physicians and other healthcare professionals utilize chronic pain assessment instruments. The survey was disseminated to patients through the organizations of several US Pain Collaborative members. Chronic pain survey respondents numbered more than 2,700, and survey results showed that the impact of chronic pain on patients' lives is not adequately, consistently, or uniformly measured. Over 90% of patients surveyed indicated changes are needed in the way healthcare professionals evaluate chronic pain.

Telling Our Story

CHRIS WODKE, TEAM CMT FOUNDER

Lots of people have seen pictures of me in my Team CMT uniform. I've been doing events since I founded the team in 2011. What many people may not know is I also wear another uniform, that of a member of the National Ski Patrol (NSP).

I've been a member of a local patrol in Milwaukee for Wisconsin for 30 years. My job is to make sure customers are skiing safely and to take care of injured skiers. I love patrolling because it gets me outside in the winter and the people on my patrol are wonderful. I've made life-long friends.

I started skiing in college and like anything athletic it took me quite some time to become a proficient skier. I took up running, swimming, cycling and rowing to become a better and stronger skier. It was the start of my life as an athlete.

It was a new experience for me since I was always the slowest runner at school and really clumsy. I tripped and fell a lot. I still do.

I founded Team CMT in 2011 to use my athletic events to raise awareness of CMT. We have over 200 athletes in 40 states and 7 countries. We have athletes doing events to raise awareness almost every weekend. It is so inspiring me to see their efforts and to hear their stories. I am so grateful for their efforts. They play a huge part in raising awareness of CMT. I am so proud of their efforts.

(Continued top of facing page)

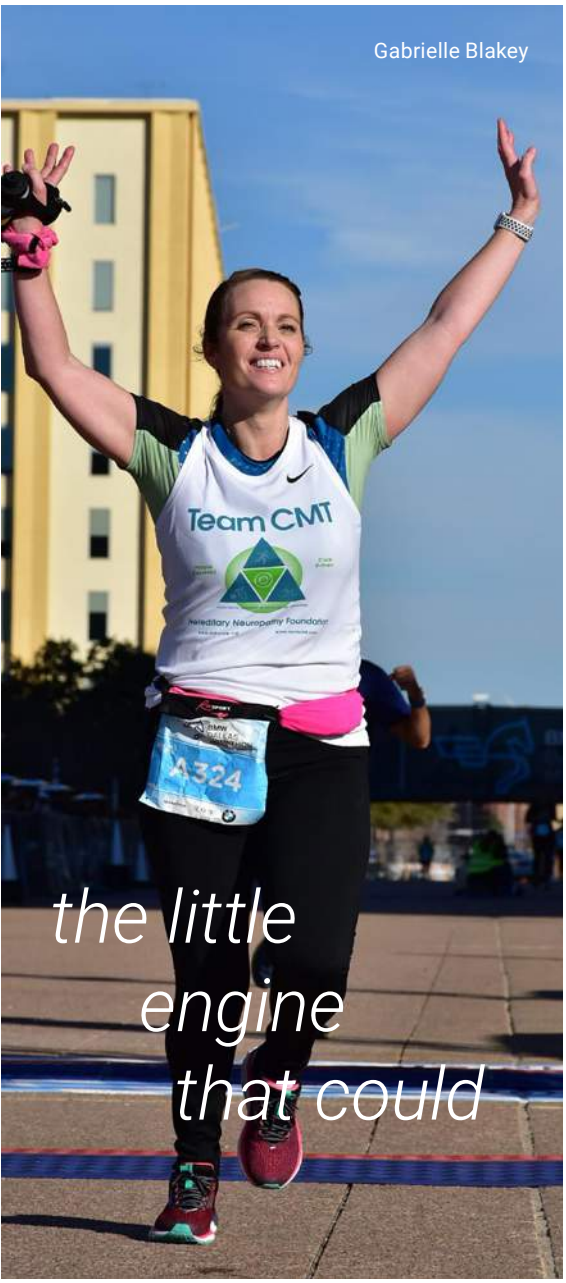
This year I got a chance to take our story to a whole new audience. The NSP has a partnership with Subaru. For a number of years, they've selected ambassador's to represent their brand. This year there were 400 applicants for the 10 ambassador positions.

To be selected I had to submit a written application online. My story of being an athlete with CMT was a large part of my application. I said I would use the car and my platform to tell the CMT story to a whole new audience. I was selected as one of two storyteller ambassadors because of my CMT. I was told the committee was intrigued by my story because it is so unusual for someone with CMT to be a patroller.

My job will be to drive my wonderful Subaru for the next year, take it to events and write about it on my blog. I will also do posts that will be appearing on run4cmt.com. Basically the same thing I've been doing for the last 9 years.

It isn't just my story, because I will be telling the story of CMT as a representative of our community. This is just another larger venue.

I hope it inspires each and everyone with CMT to tell their stories in their own way. We each have a part to play in raising awareness and raising funds to find treatments and a cure for CMT. I hope you will follow me over the next year through my blog and find your own way to tell our CMT story.



Catching Up with Team CMT Member, Gabrielle Blakey

I grew up always wanting to play sports. I was the kid that wasn't fast enough; balance and coordination were not on my side either. Always dreaming of being an athlete, I got as close as I could by assisting the coach as a statistician for Track & Field.

Growing up I knew I had CMT because my dad, his mother, and most of his siblings have it. CMT wasn't something my family talked about. It wasn't until I was 24 years old that I went to a Neurologist and was medically diagnosed.

I was working at a gym when I met a girl who was training for a Full Marathon. She came in every day to run on the treadmill and we would talk about her training. She encouraged me to run. That summer, I ran my first 5K with her. It was hard, but it felt amazing! For the first time in my life I was the one running and not writing the STATS.

Thirteen years later, on November 10th, 2019 I ran my 3rd Half Marathon in Fort Worth, TX. My personal goal was to run it under an average pace of 11 minute mile. Two of my dear friends who are amazing athletes ran with me to help pace. We were running at 9:30 avg pace. Once I hit mile 7, my knee began to hurt so bad. I've never had a pain like that before. My friend encouraged me to run through it and told me it would stop. I thought she was crazy. I was nauseated and thought for sure I had to quit. I told the girls to keep running and that I would see them at the finish line, so they went on ahead. I stopped several times, stretched, walked trying to work it out. I hit mile 9, and it became a mental game. Memories flooded my mind; how CMT has always been my stumbling block. I thought about all those with CMT who are unable to run. I thought about Chris Wodke and her story and what an amazing athlete she is. If she can do it, then maybe, just maybe I could too. I felt like "The Little Engine That Could." I decided that day that CMT does not define me, it's just a part of me.

I crossed the finish line at an average pace of 10:40 minutes/mile. Holy Cow, I did it!! I did it for me, I did it for my family, and I did it for all who have CMT. My friend was right, thankfully the knee pain subsided. I am on track to run my first Full Marathon in Dallas on December 15th.

EMBRACE THE JOURNEY AND KEEP MOVIN!

CMT-Connect Webinar: Surgery & CMT with Dr. Glenn Pfeffer

Surgery—it's a scary word filled with anxiety, apprehension and uncertainty. CMT patients are often confronted with the difficult decision to have surgery in the hopes it will help them walk easier and without pain. There are many reasons why patients opt for surgery. One of the most common is to correct foot deformities. With atrophy comes muscle irregularities, which can cause some muscles and tendons to pull on the foot unequally. Calluses, spurs, hammer toes and twisted ankles are just a few of the challenges that soon ensue. Feet become twisted, turned and extremely difficult to walk on.

So how do patients know when it's time to go under the knife? When do the potential benefits outweigh the inconvenience of casting, rehab and recovery time? More importantly, who can we turn to and trust when the stakes are so high?

Dr. Glenn Pfeffer, at IG Cedars Sinai, has been performing surgery on CMT patients for over 30 years and credits much of his passion to HNF's CEO & Founder, Allison Moore:

"*Allison Moore: really inspired me with CMT and to really forge on and to do more research. I think, certainly I'm here today in my career where I am in a large part because of her get up and go and inspiration.*"

On August 29, 2019, HNF hosted a Surgery and CMT webinar in hopes we'd be able to answer our community's questions around this important topic.

Dr. Pfeffer spoke for a full hour with informative slides, patient testimonials and a Q&A session. We are grateful to Dr. Pfeffer for sharing his time with us, and encourage anyone who has ever considered foot surgery to watch the webinar.

For more information on Dr. Pfeffer please follow him on Instagram at @charcotmarietoothsurgery

UPCOMING WEBINARS

Dating & CMT
Feb 6, 2020 @ 7pm EST

REGISTER FOR UPCOMING WEBINARS:
www.hnf-cure.org/cmt-connect

PAST WEBINARS

- 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 & Finances
- Ability360 Sports & Fitness Center
- Active Hands
- Cannabis & CBD for CMT
- CMT & Canine Companions

VIEW PAST WEBINARS:
www.hnf-cure.org/cmt-connect-webinars

CAUTION

Patients with CMT should be cautious when prescribed medication from their HCP's. The neurotoxic drug list is intended to alert patients with CMT about medications and supplements that might worsen their CMT symptoms. Please familiarize yourself with this list and share it will all your treating physicians.

One medication that HCP's prescribe often that can be toxic for CMT patients is: Cipro

DOWNLOAD THE NEUROTOXIC DRUG LIST
www.hnf-cure.org/neurotoxic-drugs

ASK THE EXPERT

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: Can CMT cause A-Fib because of the nightly severe leg cramps that constrict the muscles, which in turn cause blood clots? Or is it not possible for the leg cramps to cause blood clots?

A: Dr. Florian Thomas:
Great question. Blood forms clots in the leg when either blood flow is interrupted by a physical barrier to its flow, lack of mobility, or there is a hypercoagulability process. Muscle contractions contribute to better blood flow in the legs by pumping the blood up toward the abdomen (from legs to the thighs). It is highly unlikely that muscle cramps in isolation cause blood clot. There is no evidence to support higher prevalence or incidence of A-Fib in CMT1A. These two diseases are common and can be seen in CMT pts like the others.

Having said that, please make sure that your AFOs are not very tight blocking the blood flow in your legs.

▶ **Have a question?**
https://www.hnf-cure.org/ask-the-expert

EmBRACE It! WITH LAINIE & ESTELA

Podcast with Lainie Ishbia and Estela Lugo

Patient care is not always about keeping up with doctor's appointments and medications, it's also about how we feel in our own skin, our social interactions and relationships. In 2018, HNF hosted its third CMT Summit with a focus on behavioral health. Through multiple surveys and group discussions, we were able to gain a better understanding of the significant effect CMT can have on emotional health. Everything from dating to body image, family, career, fashion, grieving our old lives and more were addressed at this event.

As patients ourselves, we truly understand how isolating CMT can sometimes feel. We don't all have individuals in our lives who can relate to our everyday struggles. There is really nothing that compares to meeting others who are going through similar experiences and who "get it".

This summer, HNF jumped into unchartered waters to bring some "real talk" into the daily lives of patients with the "**EmBRACE It**" podcast. Lainie Ishbia of Trend-Able blog and Estela Lugo of HNF are on a mission to empower the community with real life conversations around overcoming challenges, navigating loss, bracing, family life, wellness and more.

We are four episodes in and learning the ropes as we go. Our guests so far have included: Bernadette Scarduzio, from the CMT documentary, "Bernadette", Jessica Ruiz, a disability advocate and makeup artist and Julie Stone, a CMT exercise advocate.

If you'd like to suggest any future topics or are interested in becoming a guest, contact us at estela@hnf-cure.org

"We're friends in real life, so the conversations are authentic and fun. We want our viewers and listeners to feel like they can relate to us, and that it's perfectly okay to be imperfect!"

Calling all Grandparents (and Parents)!

IRIS ADLER, GRANDPARENT OF A CMT2A PATIENT

It's hard to believe that 11 years ago my grandson, Elliot, was diagnosed with Charcot-Marie-Tooth (CMT) disease. After the age of seven, Elliot could not play soccer, ride his bike, or hike in the woods. Instead, he's endured several surgeries and is struggling today to cope with his deteriorating condition. Elliot wears braces that extend to his knees, and the loss of agility makes it nearly impossible for him to enjoy activities that most young boys and men take for granted. After meeting Allison Moore, HNF's CEO/ Founder, I partnered with the Hereditary Neuropathy Foundation and created a fund called H.E.L.P (**H**elp **E**lliot **L**ive **P**roud) specifically for research of Charcot-Marie-Tooth Type 2A. Since then, I've worked hard to raise money to cure and treat this little-known disease. It has been a successful endeavor, but we still need to do more for research and to bring drugs to market!

Allison has devoted her life to our little-known disease, which causes such distress. She has done an amazing job with new discoveries, information and making the world more aware of CMT.

We are so close, really close, to eliminating and perhaps reversing the effects of Elliot's CMT2A. What we discover for Elliot's type will benefit all the variants of CMT. So if everyone donates (even a small amount), we will be on the road to raising enough to make this a reality.

You may choose to give for research specifically for CMT2A to the H.E.L.P. Fund. **CLICK HERE:** www.hnf-cure.org/h-e-l-p-for-cmt

TO DONATE FOR THE CMT TYPE THAT AFFECTS YOUR FAMILY, CLICK HERE: www.hnf-cure.org/donate-hnf

Whatever you decide, please GIVE!

With gratitude,

Iris Adler

JOIN US for the 11th annual Charity Card Party on February 7, 2020 at Boca West Country Club.

CLICK HERE: weblink.donorperfect.com/CardPartyHELP

Dear Kristin,

We just got married. Before we spoke of having a big family, but, recently, my genetic testing showed I have CMT1a. I always thought I was just clumsy. My husband now says he doesn't want children because of the disease. I am devastated. I always dreamed of a big family.

Yours Truly,
CMT1A Patient

Dear CMT1A Patient,

I am sorry you and your husband are experiencing this. His reaction is both painful and understandable. With time and education, he may change his mind. I have never personally ever regretted my decision to have a child, but it's a very personal choice—a decision and a discussion that you and your husband should make after research and heart-to-heart conversations. Many chose to not have children out of fear of passing CMT to them. There are also options in today's world. There is preimplantation, to ensure you don't pass CMT onto your children. There is also adoption to consider. Neither are to be taken lightly, but you both should be on board with whatever decision you do make, otherwise you may wind up resenting him, and he you. Having CMT does not change who you are or what you had dreamed for in life. Let him see and understand the life you have lived is a life worth living, and maybe he'll understand that any child you bring into this world will feel exactly the same.

Congratulations on your new marriage!

Kristin Gelzinis LMSW
HNF Patient Advocate
info@hnf-cure.org

5 Simple Ways to help CMT Patients

HNF runs on passionate patients and supporters. We need you to help us continue our mission to increase awareness and accurate diagnosis of Charcot-Marie-Tooth (CMT) support patients and families with critical information to improve quality of life, and fund research that will lead to treatments and cures.

There are many ways to help support CMT research and patient programs by HNF.

1. Start a Facebook fundraiser:

1. Click **Fundraisers** in the left menu of your News Feed.
2. Click **Raise Money**.
3. Select **Nonprofit** or **Charity**.
4. Type in the **Hereditary Neuropathy Foundation** charitable organization, choose a cover photo and fill in the **fundraiser** details.
5. Click **Create**.

Make sure you add a personal message and image. Share the link with family, friends and post on social media.

2. Join Amazon Smile:

To start donating, it's super easy! Please click the link below to designate the Hereditary Neuropathy Foundation as your charity Everytime you shop on Amazon a portion will go toward the Hereditary Neuropathy Foundation. smile.amazon.com/ch/13-4137654

3. Birthday for a Cause

Start a Birthday Fundraiser! It's simple and a great way to spread awareness to friends and family in just minutes.

1. Click on link below.
2. Enter your name and address and submit.
3. Look for a follow-up email from me that has a link to make your own fundraising page.
4. Click the link and look for yellow words on the page "Create My Own Fundraising Page" and click.
5. Start making your page! Make sure you add a personal message and image. Share the link to family, friends and post on social media.

Click here to make your Birthday Fundraiser page: www.hnf-cure.org/birthday-donation/

4. Launch a Letter Writing Campaign

Launch a letter writing campaign and raise funds for CMT research.

HNF has created a template that you can use and customize to your form of CMT.

All donations will go towards the research of your choice.

If you have any questions please email: courtney@hnf-cure.org

5. Sign up for the RoundUp App

HNF has partnered with an app called RoundUp. This app allows users to donate their change to support CMT research by rounding up to the next dollar from credit or debit card purchases – all automatically and without hassle. You can even cap the maximum donation amount in a given month.

Sign up today! www.hnf-cure.org/roundup-app

Or simply download the app or use the web version at roundupapp.com. You'll be able to create an account and choose HNF when prompted to select the organization you will support.

Visit HNF's Get Involved page for more ways to get involved! www.hnf-cure.org/get-involved-for-charcot-marie-tooth-awareness-month/

HNF ON THE ROAD:

American Association of Neuromuscular and Electrodiagnostic Medicine (AANEM) Annual Meeting

We hear time after time from frustrated CMT patients across the country, "My doctor doesn't know much about CMT. How do I know I'm getting the best treatment?" HNF understands the impact and scale of this challenge. It's what drives us to connect with neurologists, neuromuscular physicians, technologists and other professionals every chance we get.

"There are hundreds of neuromuscular diseases out there affecting many people," said American Neuromuscular Foundation Director, Shelly Jones. "Supporters typically only hear about a few, like ALS or SMA. However this approach leaves those other diseases without exposure."

CMT has been one of those unexposed diseases for many decades, but our mission is to change that. For over 9 years, HNF has been representing the CMT community by attending, showcasing and presenting posters, running workshops on Patient Reported Outcomes at the American Association of Neuromuscular and Electrodiagnostic Medicine (AANEM) Annual Meeting, and finally collaborating on executing two Continuing Education Courses through government and industry support.

This year's meeting was held in Austin, TX. October 16-19 with more than a thousand attendees working in the fields of neuromuscular (NM), musculoskeletal (MSK), and electrodiagnostic (EDX) medicine. The focus this year was the present and future role of technology in treating patients with neuromuscular diseases, according to AANEM and ANF President Anthony Chiodo, MD. "Technological advances are rapidly occurring in the areas of robotics,

computerization, mobility aids, and others," Dr. Chiodo said. "Each of our speakers will share how they believe technology will translate to improved patient care and the continued cultivation of future technological breakthroughs."

Important data posters, captured from HNF's Global Registry for Inherited Neuropathy, GRIN were presented by HNF to better provide attendees with the CMT patient perspective and what matters most to them.

- **Charcot-Marie- Tooth 1A and Impaired Patient Mobility:** Expressions, Remedies and Impact on Quality of Life
- **The Path To Diagnosing Charcot-Marie-Tooth Disease:** The Patient Experience
- **Qualitative and Quantitative Voice Activation Technology captures Patient Reported Outcomes to inform the Food & Drug Administration on Charcot-Marie-Tooth Disease**

The HNF booth displayed the latest CMT materials and provided attendees with resources for their practices as well as their patients. Physicians were encouraged to take the free credited CMT Course (https://bit.ly/2EP1Yye) developed in 2010 and the Assessment Test (https://bit.ly/34QKN9Y), recently developed in 2018.

Left to right:
Courtney Hollett, Allison Moore and Estela Lugo

"We want physicians to have as many tools in their pocket to better treat the CMT community. We want patients to leave their appointments feeling empowered by the work HNF does on their behalf and connected to a large network of meaningful resources."

– Estela Lugo,
Medical Outreach Manager.

Hope for the Future for Owen

HOPE LOPEZ IS A SINGLE MOTHER OF 2 WHOSE SON OWEN HAS CMT4 GDAP1

Q. CAN YOU TELL US A LITTLE ABOUT YOURSELF?

My name is Hope Lopez. I'm 28 years old and a single mother of three amazing boys, Owen (6), Oliver (21mo.), and Remington (2mo). I have moved around quite a bit to be where I have the most support for my kids. Currently, that has brought me to the small town of Burlington, IL. I live with my father, Raul, and my children. My father has always been my biggest support system, and within the last year and a half, I was blessed with meeting Marty and Jose. They are a gay couple who have become a tremendous help and support for me and my kids—caring for them while I work, coming along to doctors' appointments, and helping with Owen's school. They have become a part of my family. I work as an EMT Basic and have been employed at AMT for four years. My medical background has been a big help in understanding and dealing with hospitals, nurses, medical procedures, etc.

Q. WHO IN YOUR FAMILY HAS CMT4 GDAP1? WHEN WERE THEY FIRST DIAGNOSED? WHAT TYPE OF SYMPTOMS DID YOU FIRST NOTICE?

There is no history of CMT4 GDAP1 in my family history. My son Owen is the first to be diagnosed after foregoing years of tests and seeing different doctors, many of which began treating his symptoms before knowing the cause. Owen was born with a congenital heart defect, HLHS, so he had been receiving speech, developmental, and physical therapies since he was six months old. His larger milestones were the easiest to notice first as he was delayed in reaching them. Around 15 months, we noticed that he wasn't walking and the physical therapist noticed that he had low muscle tone in his lower legs and drop foot. She had suggested AFOs, a vest to support his torso and theratog shorts. As time went on, we began to notice other things such as his toes curling over, sensitivities to different textures and temperatures, and then, later, his hands began to contract. He was finally diagnosed on October 7th of 2019 after two different types of genetic testing.

Q. HOW LONG DID IT TAKE TO BE DIAGNOSED?

Because Owen was born with a congenital heart defect, most of his delays were thought to be heart-related at first. We had been referred to a neurologist when Owen was about three years old. He had many tests that were all coming back normal. After his third surgery, we noticed weakness in his hands, and were then referred to a surgical neurologist who diagnosed Owen with neuropathy and suggested that he get genetic testing to see what kind of neuropathy he had. It took two rounds of genetic testing and more than 18 months to get the diagnosis.

Q. WHO DIAGNOSED HIM?

Dr Rao, Surgical Neurologist at Lurie Children's Hospital in Chicago, diagnosed Owen with GDAP1 after genetic testing.

Q. HAD YOU EVER HEARD OF CMT BEFORE THIS?

I had never heard of CMT before this.

Q. WHAT DID YOUR DR. TELL YOU TO EXPECT? WHAT ADVICE/RESOURCES DID THEY PROVIDE?

Dr. Rao informed me that Owen had a type of neuropathy that was very aggressive and was progressing extremely rapidly. This was prior to the genetic testing. He said to expect it to get worse and that there would be no cure. He did not give us any resources. He only had us continue physical therapy which Owen was already doing. After he was diagnosed with GDAP1, Dr. Rao suggested Owen be included in clinical trials for GDAP1. He also said to register Owen in his neuropathy clinic starting in January 2020.

Q. HOW DID YOU FIRST LEARN OF HNF?

After finding out about Owen's diagnosis and not receiving much information on it, I started doing my own research online. I had found that GDAP1 was linked to CMT and started doing more research on CMT, as there is not much information to go by on just GDAP1 alone. Once I started the research on CMT, I began looking for support and informational groups on Facebook. I had shared my concerns about Owen's new diagnosis on one of the CMT pages and asked if anyone had the same form of CMT and, if so, would anyone be able to share information with me. From there, someone tagged Estela Lugo in the comments. I reached out to Estela and saw that she was involved with HNF and started looking into it.

Owen Lopez

Q. WHAT ARE SOME OF THE BIGGEST STRUGGLES FOR YOUR SON?

Owen has very little muscle tone below his knee in both legs and the same in his hands and wrists. He requires support in walking and wears AFOs. In addition, his heart condition affects his stamina and stairs are particularly hard for him. He is not able to open his hands all the way. They are always in a semi-clenched position, therefore making it difficult for him to write, feed and dress himself, among other issues. He also has sensory issues, which vary day to day. Although Owen wants to be independent, he is still very dependent, and finds himself aggravated and frustrated when there are things he wants to be able to do but just can't.

Q. HOW DOES CMT AFFECT YOUR FAMILY?

Owen has therapy in school and two/three times a week at a Lurie Children's clinic, which is more than an hour away. This is a big time constraint. Owen has to miss school frequently, and we often have to rearrange our work schedules to accommodate this crucial therapy. Owen can't keep up with his peers on a physical level at school and has difficulty going up the stairs at home and getting on and off his school bus. Almost everything takes a bit longer for Owen. The additional time and attention Owen requires does put a financial burden on the family but, with God's help and those who love and support us, we always get by. We love Owen immensely and gladly give him all we can to help him to thrive!

Q. WHAT ARE YOUR HOPES FOR THE FUTURE?

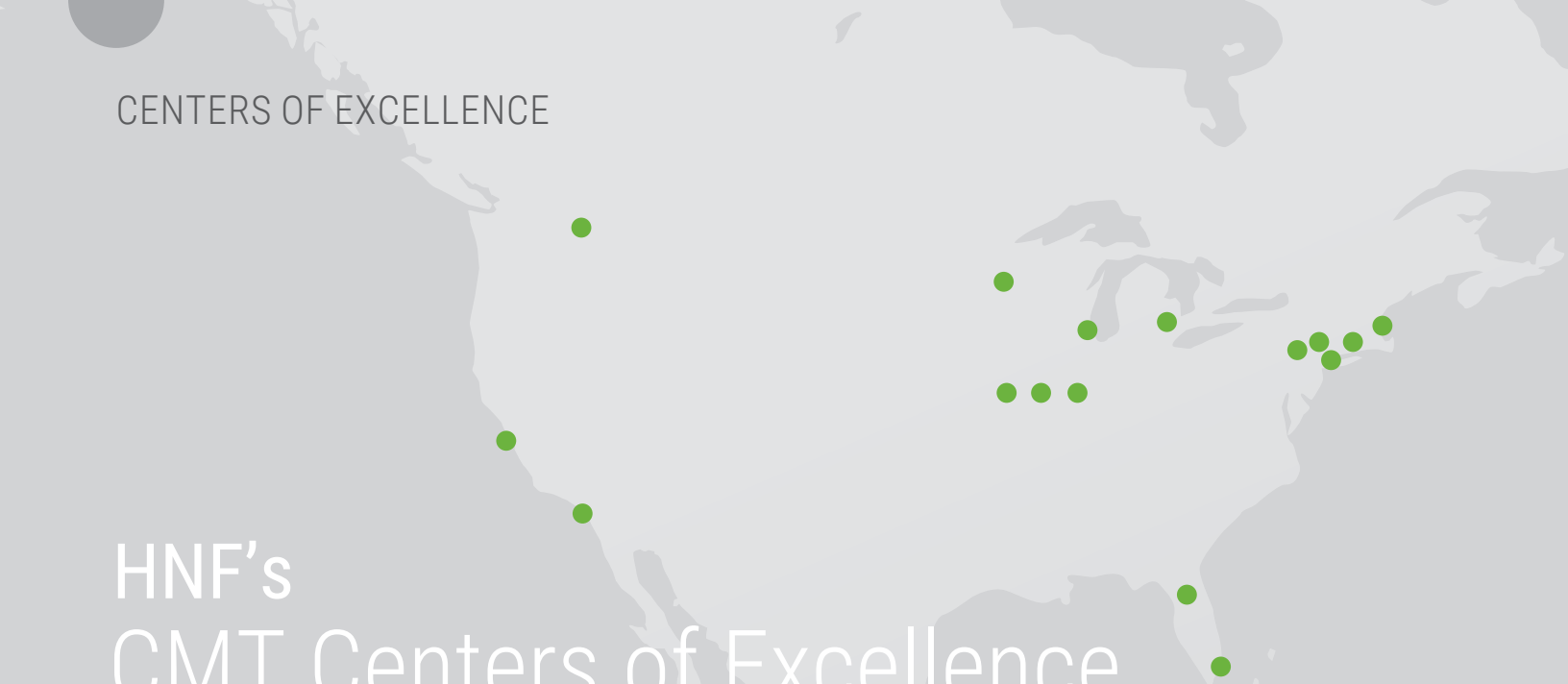
Our hopes for the future are that a cure is found and that Owen can lead a "normal" life.

Therapeutic Research In Accelerated Discovery (TRIAD):

A gene therapy is within our reach. GDAP1 (CMT4A) is an autosomal recessive loss of function disorder which our scientific advisory board and CMT research collaborators have identified as a type of CMT that has the potential to be cured with gene therapy. HNF is committed and needs your help!

Calling all GDAP1 patients to support our gene therapy program. HNF is developing laboratory GDAP1 animals and Induced Pluripotent Stem Cells (IPSc) human cell models. This first phase of the project will take approximately 6 months to complete.

TO SUPPORT, PLEASE DONATE: www.hnf-cure.org/cmt4-gdap-gene-therapy
FOR MORE, CLICK HERE! www.hnf-cure.org/cmt4-gdap-gene-therapy
TO PARTICIPATE IN THE RESEARCH, CONTACT ALLISON MOORE:
allison@hnf-cure.org



HNF's CMT Centers of Excellence

The national network of HNF-designated Centers of Excellence (COE) provides patients with resources to find hubs of expertise in caring for and treating CMT, as well as locations where CMT research is being conducted. Our primary goal 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

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
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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

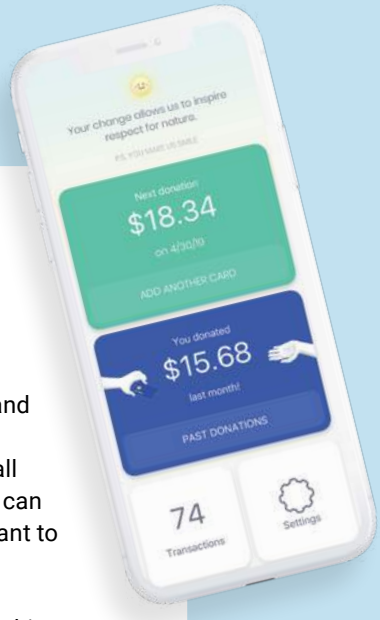
NEW YORK

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. You will be able to create an account and choose us when prompted to select the organization you will support. Also, we would love for you to spread the word to other individuals who may be interested!

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

Thanks,
Courtney

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Celebrating CMT September Awareness Month

CMT Awareness Month 2019

HNF put its Movement is Medicine™ program in play by hosting TEAM CMT Spin Events on the west and east coast. A big shout goes out to Tara Emerson, CMT patient and exercise enthusiast and Revolution Fitness for donating the space in Santa Monica, California.

We are so grateful to the PIKE fraternity at Boston University and Acceleron Pharma for sponsoring the 2 hour TEAM CMT Spin at Handlebar in Boston, MA.

Both events exceeded our expectations with over 60 people participating and all together we raised $15,000!

To date, HNF has sponsored 10 spin events and has raised over $500,000 for CMT Research. To keep the momentum going, we are counting on our HNF members to consider hosting their own TEAM CMT event. It's turn key and we will be by your side all the way.

To host a TEAM CMT Spin Event
contact allison@hnf-cure.org

PHARNEXT

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