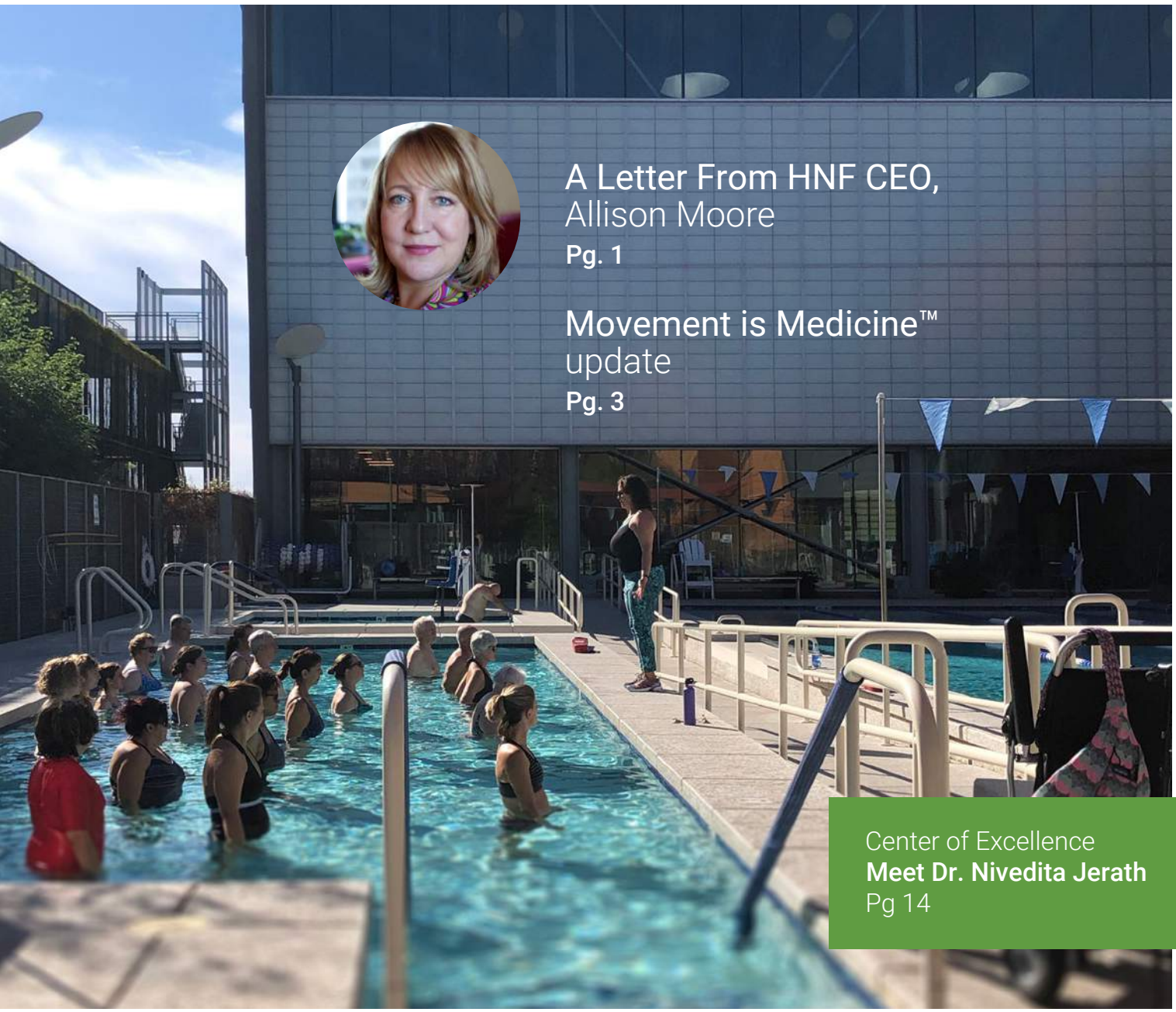
HEREDITARY NEUROPATHY FOUNDATION

WINTER 2020

cmtupdate



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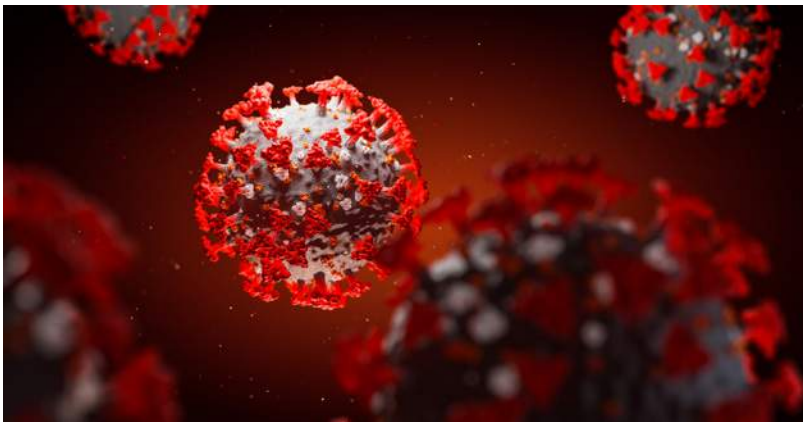
BolleDesign.com

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

www.hnf-cure.org

CMT UPDATE
WINTER 2020



YOUR SOURCE FOR CMT & COVID-19

CLICK HERE:
www.hnf-cure.org/covid19

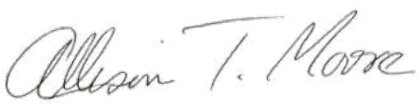
Dear Friends:

When I originally wrote my "Letter From The CEO" for our Winter 2020 CMT Update several weeks ago, the world was quite a different place. The threat of Coronavirus still seemed distant — was something that was happening "somewhere else" — and the HNF team was busy planning four Movement is Medicine™ one-day regional summits, plus our big two-day event in Phoenix in November, as well as multiple exercise-related fundraisers throughout the country. All those plans and events came to a screeching halt with the rapid escalation of COVID-19 in this country and the accompanying governmental response, which has effectively restricted travel and shut down any gathering over 50 people.

While these restrictions will impact our ability to raise money for our programs in the short term, our priority as an organization has and always will be the health and safety of our members. We encourage you all to follow all guidelines released by the CDC and other government agencies to keep you and your family as safe as possible. While there is no evidence that people with CMT Disease as a whole are at greater risk than the general population if they are infected with COVID-19, those patients with a compromised immune system or comorbidities such as respiratory issues should be extra vigilant by avoiding high risk situations and developing good preventative hygiene habits.

Even though we are being encouraged to practice "social distancing" in our everyday lives, we at HNF will always be here to support our community as we all navigate this crisis together. I encourage you all to join our Inspire community, if you are not already a member, and to participate in our webinars when you are able to. We need each other, now more than ever!

All the best,



Allison T. Moore, Founder/CEO, Hereditary Neuropathy Foundation

PS: HNF relies on the generosity of our community to help support our programs and research. With most of our fundraisers put on hold for the moment, there are many ways to help us reach our goals...please take a look here! **WWW.HELP4CMT.ORG**

MOVEMENT IS MEDICINE™ SUMMIT 2020

Fingers Crossed!

The Movement is Medicine™ Summit is tentatively scheduled November 13th & 14th at Ability360 in Phoenix, AZ, subject to COVID-19 travel and gathering restrictions. We remain cautiously optimistic about being able to hold this event.

UPDATE 3/16/20: As of now, most local and state governments are restricting gatherings to no more than 50 individuals, and people are being encouraged to stay in place, limit non-essential travel, and avoid large crowds. It is possible that these restrictions will start to ease in the next several months. With that in mind, we are keeping the Movement is Medicine™ Summit 2020 on our calendar for now, with the hope that we will still be able to hold this important event for our community in November.

Last November, HNF made history by holding the first annual Movement is Medicine™ Summit at the universally accessible Ability360 Sports and Fitness Center in Phoenix, AZ. Attendees were treated to two days of inspirational sessions, hands-on exercise classes, and informational break-outs on bracing, nutrition, clinical trials and more!

Kicked-off by a motivational keynote by Mike Studer, PT, MHS, NCS, CEEAA, CWT, CSST, attendees were able to create their own agenda by selecting sessions or topics that were of most interest to them. Attendee favorites included Aqua Yoga with HNF's own Bernadette Scarduzio, Core Strength with Tara Emerson, Vector Training with Kayezen Fitness, and adaptive rock climbing. Attendees were also able to get one-on-one surgical consultations with leading CMT surgeon Dr. Glenn Pfeffer and one-one-on discussions with Paralympic swimmer Jamal Hill and keynote speaker Mike Studer.

One attendee said, "I had never been in a room where I felt so safe in my own skin. I felt safe because I knew everyone around me had awareness of CMT and it is something that affects all of us in many different ways. The ripple effect a gathering like this does for CMT is infinite!"

We will hopefully be able to build on this momentum with Movement is Medicine™ Summit 2020, once again at Ability360 in Phoenix, Arizona on November 13th &14th. Based on attendee feedback from last year's summit — which was overwhelmingly positive — we are planning to make this summit even better with more sessions, breakouts and programs specially designed for patients with CMT.

Because of the current restrictions of travel and group gatherings, we are not selling tickets to the Summit at this time. You can, however, join our free "pre-registration" list to first get a crack at purchasing tickets should we go forward with the event sign up.

PRE-REGISTER! www.hnf-cure.org/movement-is-medicine-summit-2020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OPEN CALL FOR MOVEMENT IS MEDICINE™ SUMMIT 2020 PARTICIPATION

After the overwhelming success of our Movement is Medicine™ Summit 2019, HNF is calling upon YOU to help us make this year's event even better!

HNF is calling all CMT patients, family, caregivers and healthcare providers and others to submit one or all of the following for the Movement is Medicine™ Summit on November 13-14, 2020:

- Proposed topics and/or speakers for workshops
- Ideas for adaptive exercise classes
- Nominations for class instructors
- Contact information for potential sponsors

Please submit your ideas to:

cmtsummit@hnf-cure.org

No later than April 30, 2020.

From the submissions, we will randomly select a winner to receive a $50 Amazon gift card.

Please be sure to include:

- Your full name and email address
- A complete description of your idea and/or complete contact information for the person/company you are nominating

TRIAD
THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY

GLOBAL REGISTRY FOR INHERITED NEUROPATHIES

The Global Registry for Inherited Neuropathies (GRIN) powers the Hereditary Neuropathy Foundation's patient-centered research and drug discovery initiatives.

Leading Charcot-Marie-Tooth advocacy organization's innovative platform plays a critical role in its mission.

BY **ALLISON T. MOORE,** HNF FOUNDER AND CEO

Our registry, GRIN, has been central to our patient-centered research initiatives for over 6 years. The ability to have real-time access to important patient reported natural history data has been a game changer for HNF's research, industry and academic partners. Originally hosted using 3rd party software, in 2019 HNF migrated GRIN onto our own IRB approved, HIPAA compliant platform. Built to our own specifications, GRIN allows us to quickly analyze data to uncover insightful correlations between genotypes, phenotypes, comorbidities and other important patient data.

GRIN has played a critical role in identifying important issues related to the CMT patient experience, revealing new areas to explore and research. When pain was noted by 65% of patients with inherited neuropathies, HNF quickly sprang into action, hosting a dedicated summit on patient pain in 2017, as well as advocating for pain scales to be considered as a secondary endpoint for clinical trials. "Without real time access to this critical data point, we wouldn't have been able to demonstrate the importance of this issue to our industry and research partners," says Joy Aldrich, HNF's Patient Advocacy Director. "**GRIN** also helped us effectively convey the CMT patient's disease experience for our Externally-led, Patient Focused Drug Discovery meeting with the FDA in 2018."

GRIN has also facilitated recruitment for the latest clinical trials by pinpointing ideal candidates with speed and accuracy. Future innovations for the platform include an interactive patient portal, the integration of de-identified data with other partner consortiums, and the collection of longitudinal patient data. **GRIN** also supports the CMT&Me app, sponsored by Pharnext, by identifying patients that are willing to participate in that important digital lifestyle study.

"GRIN plays an important role for clinical trial recruitment, facilitating the possibility of enrollment for ideal candidates."

Xavier Paoli, VP & Chief Commercial Officer, Pharnext

To participate and accelerate research, join GRIN. www.hnf-cure.org/registry

Contact Allison Moore allison@hnf-cure.org

TRIAD
THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY

$61,000 Raised to Support Pediatric CMT Trials

Last summer HNF teamed up with the Penn Medicine Orphan Disease Center (ODC) for the Million Dollar Bike Ride (MDBR) in Philadelphia. Our fundraising goal of over $30k was achieved and matched by the ODC, granting HNF over $60k to allocate to CMT research. Thanks to our generous sponsors and riders!

After evaluating many different potential avenues, HNF chose to direct the research funds to a very underserved, yet vital area — pediatric clinical trials. "We have very promising developments taking place in pharma and genetic therapy research right now, but without the collection of more pediatric data to measure outcomes and efficacy, these treatments will never make it to our community."

When it comes to CMT in children, it's even more important to get treatments out swiftly and halt/reverse progression early on.

"It is imperative to shed more light on pediatrics and the urgency of delivering treatments to these patients and families. There are a million moving parts to produce successful clinical trials. It takes lots of work behind the scenes that few people are aware of unless they are deep in the trenches. We're confident we'll be able to significantly alleviate future hurdles and add significant knowledge and resources. When it comes to CMT in children, it's even more important to get treatments out swiftly and halt/reverse progression early on" Allison Moore, HNF Founder and CEO.

HNF is pleased to provide funding to continue the promising research being conducted at the prestigious Connecticut Children's Gait Lab. The project will be overseen by Director of Research and Education, Sylvia Ounpuu, MSC, and Gyula Acsadi, MD, PhD, Division Head of Neurology. Both investigators are a part of the Inherited Neuropathy Consortium, as is HNF.

The study will measure abnormalities in gait function using comprehensive gait analysis techniques and wearable activity monitoring and then compare them to CMTPedS scores. The research will also compare the changes in these parameters over time and study the relationship between the patients' measureable gait function and a promising biomarker, neurofilament light chain (NfL) level. The information gained from these investigations will support not only the understanding of the natural history of gait and community function, but also clinical trial readiness for new treatments. "To conduct a successful therapeutic trial for children and youth with CMT, a detailed understanding of the natural history of walking decline is needed, along with predictable benchmarks to support the development and translation of emerging disease-modifying therapies," Sylvia Ounpuu, MSc.

CALLING ALL FAMILIES AFFECTED BY GDAP1 (CMT4)

HNF is currently looking for families to join our emerging gene therapy study for this rate type of CMT.

EMAIL Estela@hnf-cure.org to join our April research update call!

Meet the Kohler Family: Alana has GDAP1

Q: CAN YOU TELL US A LITTLE ABOUT YOURSELVES?

We live in Lino Lakes, Minnesota. Our family includes Luke, Robin, Landon (13), Alana (11) and our dogs Mylie and Citori. Luke is a project manager and Robin is a radiologic technologist.

Q: WHO IN YOUR FAMILY HAS GDAP1 (CMT4)? WHEN WERE THEY FIRST DIAGNOSED? WHAT SYMPTOMS DID YOU FIRST NOTICE?

Alana has GDAP1 (CMT4). We started noticing symptoms around age 3 with frequent trips and falls. It was also apparent that keeping up with her peers was challenging. She's been sporting AFOs since the young age of 3.

Q: HOW LONG DID IT TAKE TO BE DIAGNOSED?

Alana started showing symptoms at age 3. We were given a diagnosis of CMT when she was 5, and then we received a genetic diagnosis of GDAP1 (CMT4) shortly after her 9th birthday.

Q: WHO DIAGNOSED HER? (TYPE OF DOCTOR)

Neuromuscular Neurologist.

Q: HAD YOU EVER HEARD OF CMT BEFORE THIS?

Never.

Q: WHAT DID YOUR DOCTOR TELL YOU TO EXPECT? WHAT ADVICE/ RESOURCES DID THEY PROVIDE?

The doctor informed us that GDAP1 (CMT4) is a genetic degenerative disease and it's considered moderate to severe on the spectrum. We were given a few websites to check out and referred to the MDA.

Q: HOW DID YOU FIRST LEARN OF HNF?

Once we learned that Alana had GDAP1 (CMT4), we searched the internet and joined every Facebook support group we could find relating to CMT. We learned of HNF and their commitment to support a cure for CMT. In September we learned of the GDAP1 (CMT4) gene therapy project through the online newsletter.

Q: WHAT ARE SOME OF THE BIGGEST STRUGGLES FOR YOUR DAUGHTER?

The list is endless, but some of the most difficult moments in our lives are shoe shopping, trips and falls and the ask of "Dad/Mom, why can't I be like the other girls and just blend in?" We always try to refer to the movie Wonder and tell her, "it's hard to blend in when you were born to stand out."

Q: HOW DOES CMT AFFECT YOUR FAMILY?

CMT affects us daily, as we always try to plan our family activities with Alana's abilities in mind. We have constant battles with AFOs, walking, falling, muscle fatigue, and emotional stress due to CMT and middle school. Multiple surgeries, numerous doctor appointments and weekly PT appointments have made us a stronger and closer family.

Q: WHAT ARE YOUR HOPES FOR THE FUTURE?

Our hopes and prayers for the future include:

- Alana to walk without braces
- No progression of her symptoms
- A cure for all types of CMT and especially GDAP1

DONATE:
weblink.donorperfect.com/GDAP1

Number 34:
Rishi

Rishi Sharma CMT 2A
Wheelchair Rugby

I am Rishi and I have CMT2A. I was first diagnosed at two years-old. When my parents asked what could be done, they got the famous and horrific answer so many of us have gotten, “nothing can be done and there is no treatment. The disease is progressive and will only get worse to the point where he may end up in a wheelchair” (mic drop moment). I look back on that and kind of laugh at that being considered the “worst” prognosis. There are far worse outcomes in my opinion. The doctors were right about the progression aspect as I started to experience foot drop, hand and wrist weakness, and fatigue as I got older. By age 13, I was fully using a wheelchair to get around. This transition was a hard one as people did not understand (including myself for a long time) why I went from walking to being in a wheelchair. I got questions like “What happened?” or “Can you still walk?” from strangers, friends, and even family members. These questions were hard to answer and they left me with emotions like anger, sadness, worry, and depression. I was in denial of my disease for a long time.

It wasn’t until 16 years of age that I started embracing my disease and the wheelchair; however,I needed a new one. Just like AFOs, a well-fitted wheelchair that has everything you need can be life changing for the user. I got a TiLite ZR made exactly to my specifications and it has made my life far easier and made me more independent. I had to look at the positives of being in a wheelchair, like never having to worry about losing my balance, getting a great arm workout everyday pushing myself, and not having to wear AFOs (which I hated) everyday just to be able to get around. The wheelchair also opened up

the possibility of me being an athlete, which I never thought would be an option for me. Trying wheelchair sports changed my life for the better.

I started off playing wheelchair basketball and I realized it wasn't for me. Then I found wheelchair rugby. What attracted me to the sport was that all athletes must have deficiencies in three or four of their limbs to be able to participate. This put me at an equal playing field for once in my life.

It's a hardcore activity and I've met many great people with similar or the exact same disability as me who also play this sport. What intrigued me the most about this sport was just how many Paralympic athletes there were who played wheelchair rugby for their country's team and have CMT. **Almost every wheelchair rugby team that qualifies for the Olympics has at least one player on the team that has CMT or an inherited neuropathy.** I hope and train for the day to be on the U.S. wheelchair rugby team. With this sport comes a lot of training. I use specialized lifting hooks (which have been game changers for me) that I use at the gym to lift weights and do cable exercises. I truly believe the only way to currently slow down/stop the progression of this disease (in my opinion) is exercising and eating right.

I have shared my passion for wheelchair rugby and weightlifting with you, but there is one more passion that tops both of these. That passion is to find a treatment/cure for CMT. I dove deep into the world of neurodegenerative diseases and have found a common trend amongst most of them, there is no treatment. This is starting to change however with the discovery of highly specialized gene therapy. The big issue with CMT is there are so many different genes that can cause this disease and each subtype needs a different form of gene therapy to correct whatever the error is in the genome. At the bare MINIMUM I want to see a treatment that halts the progression of CMT2A and other subtypes.

I am glad to see non-profits like the Hereditary Neuropathy Foundation (HNF) team up with the FDA, researchers, biotech companies, and investors to push effective, life-changing treatments into the clinic. I am extremely optimistic for the future and want to continue to work with the HNF on achieving the goal of finding a treatment/cure for all forms of CMT.

Learn more about the rules for Wheelchair Rugby:
https://www.hnf-cure.org/cmt-athletes/wheelchair-rugby

Donate to CMT2A:
https://www.hnf-cure.org/donation-form

Dear Lainie @ Trend-Able,

Thank you so much for all your shoe recommendations for AFO wearers and all your posts in general. As a 50ish woman with CMT who has hid it most of my life, your blog has helped me to feel less alone. I have laughed out loud reading many of your articles and shared them with family and friends so that they can try to understand what I go through.

I have a question about bras. I still want "the girls" to look good under clothing but putting on a bra with my CMT hands is proving to be more and more difficult. Not only is the clasp hard for me to do, but the elastic digs into my skin & leaves marks. Do you have any suggestions?

Thank you,
Marilyn M.

Dear Marilyn,

Thank you so much for following Trend-Able and for your sweet comments. I'm so glad you find Trend-Able's blogs relatable and helpful! You are also not alone in the bra problem! Like you, I want to look and feel my best and my "girls" need some serious help.

Unfortunately, most of the "adaptive" bras being sold today are matronly looking and aimed at post-surgery patients and nursing mothers. But, there are some other options:

If you're able to lift your arms in order to put a bra over your head, I've found this bralette by Spanx and one from Soma to be both comfortable and supportive. To get them on more easily (and prevent the chafing you mentioned), try sprinkling talcum powder on your body first. Another easy hack is to apply a non scented deodorant or anti chafing stick under your bra line and the folds of your skin.

You can also purchase a magnetic button front closure bra (https://amzn.to/2TZjBma) or magnetic clasp extenders (https://amzn.to/3386Klq) to use on bras that you already own to replace the tricky hook and eye clasps. Finally, when you have a weak pincher grip, fabric loops can be sewn on the inside of bras and other types of clothing to allow your thumbs to fit inside and help with getting items on and off.

Xoxo
Lainie

Lainie Ishbia, MSW
Disability Speaker & Blogger
Trend-ABLE

Spinning 4 CMT: My CMT Story

MARC DAIGLE, 34 YEARS-OLD **HOMETOWN:** ST. CHARLES, MO **CURRENTLY:** NEW YORK CITY

I have been an active person my entire life. I've completed 6 marathons, 2 ironmans, 2 half ironmans and multiple other races. In 2016 while I was training for the NYC half marathon, I rolled my ankle badly and took a hard fall. Ever since that training day, I haven't been able to run the same. It wasn't until 2017 that doctors told me I had CMT. We are still trying to figure out the type, which is extremely frustrating.

Looking back, I've always shown signs of CMT. I frequently sprained my ankles, tripped over things and my hips gave out quickly while running races.

Fast twitch movements affect me the most or prolonged periods of activity. Running, walking, and jumping are the hardest. After my fall in 2017, I stopped competing in races because I didn't think I should; I couldn't run fast anymore and I didn't know how pushing through in the run would affect my body, currently or long term. After several years, I was determined to race again. I modified my training and my running gait to help me complete the NYC Triathlon in 2018. In 2019, I completed Ironman Lake Placid, which is a 2.4 mile swim, 112 mile bike and 26.2 mile run.

My symptoms now include nerve pain, nerve/muscle "popping" and twitching, and constant fatigue/discomfort.

I have been an indoor cycling instructor at Flywheel Sports since March 2016. My CMT ramped up during training. Spinning helps my CMT by allowing me to continue to move my body and muscles with low impact. Biking and swimming make me feel strong and confident.

The biggest adaptation is listening to my body. Learning what activities are hard and which ones are more doable. I'm also constantly learning and relearning what my body is capable of doing. I'm able to know the intensity of exercises and when my CMT will pass the point of no return in regards to leg and hip muscle movement. Examples include: altering my gait while running, knowing how much resistance and speed I can do safely on the bike, and completely eliminating certain exercises.

Overcoming insecurity is a constant battle and something I still work with. In the fitness industry, I was trying constantly

Fast twitch movements affect me the most or prolonged periods of activity. Running, walking, and jumping are the hardest.

to keep up with my peers, but this was not possible...in terms of competing. But the more I showed up and just participated, the more confident I became. That's why finishing those races was so important to me. Opening up to friends, family, and clients also helped me a TON. It made me feel seen and I learned how inspiring I can be.

Movement is Medicine™ is EVERYTHING to me. We literally must use our bodies and move to stay healthy and strong. If not, we will all fall to additional atrophy and we can't let that happen. Movement makes me smile. Movement makes me laugh. Movement gives me confidence. Movement makes me LIVE!

I am extremely excited to lead the Team CMT Spin New York City on Saturday, May 2nd. What a perfect way to get together, raise awareness, and build community. I'll see you in the stadium.

Register for Team CMT Spin New York City:
https://bit.ly/336cXOP

Cheers to moving our bodies.
–Marc

Inspire/CMT App and Hot Topics in Our Community

There is now an iOS App to stay connected to our CMT community using your Apple mobile device!

Here are a few of the Hot Topics:

1. Is CBD helpful?
2. Standing without bending knees
3. Has anyone had foot/ankle surgery to correct a CMT foot?
4. Hip replacement and CMT
5. How has a pet affected your life?

DOWNLOAD THE APP TODAY, OR JOIN US ONLINE:
www.inspire.com/groups/charcot-marie-tooth-cmt

Swipe Right for CMT

This February, HNF continued the conversation on CMT and dating with a new EmBrace-It podcast and CMT-Connect webinar. We heard from both the male and female perspectives on finding love with a disability from Dating Coach on Wheels, Amin Lakhani, as well as from HNF's Estela Lugo and Trend-ABLE's Lainie Ishbia. Check out both episodes for online dating tips, red flags to avoid, comedic screenshots and more!

EmBrace-It Podcast: Episode #5
hnf-cure.org/podcasts

CMT Connect: Dating & CMT
https://bit.ly/2TBm2fA

UPCOMING WEBINARS

You asked...we listened!

Get your genetic testing questions answered live!

Join us with genetic counselor Andrea Paal from Quest Diagnostics. April Date TBD

REGISTER AND SUBMIT YOUR QUESTIONS
www.hnf-cure.org/cmt-connect

PAST WEBINARS

- 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 & 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

This medication that HCP's prescribe often can be toxic for CMT patients.

Levofloxacin. An antibiotic commonly prescribed for bacterial infections like middle ear infections, strep throat, and pneumonia.

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: I have symptoms consistent with CMT, but because my symptoms are asymmetrical my doctor said I don't have CMT because CMT is always symmetrical. Is this true?

I have left enlarged sciatic nerve segment (3cm diameter), left peroneal neuropathy, left high arch foot with slight hammertoes, mild scoliosis, and my right foot has a slightly high arch and slight decrease in muscle tone in the small muscle on the side of the right foot, but the right side seems pretty much normal.

A: Dr. Florian Thomas:
While typically CMT presents with fair symmetry, this is not always 100%. One CMT related condition in which symmetry is not expected is HNPP (hereditary neuropathy with liability to pressure palsy). With all other forms of CMT there should be symptoms & signs of neurological problems on both sides of the body, even when there is asymmetry. Electrodiagnostic studies and genetic testing should be considered. However many conditions can result in the clinical features you describe. I suggest you take all existing studies to the neuromuscular specialists at an academic medical center for a 2nd opinion.

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

Left to right:

Laura Patrick, Jean Macaluso, Carlos Hernandez, Nivedita Jerath, Allison Moore, Ashley Hayden, and Craig Brubaker.

Meet Dr. Nivedita Jerath, MD, MS

AdventHealth Neuroscience Institute Center of Excellence

Dr. Nivedita Jerath

Q: TELL US ABOUT YOURSELF?

I love my patients and treat them like friends and family.

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

Q: WHY DID YOU CHOOSE CMT AS YOUR FOCUS OF STUDY AND RESEARCH?

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

Q: WHY IS CMT YOUR PASSION?

For many reasons! I love finding new genes and mutations that cause peripheral neuropathies. Genetics is a new field of neurology, in which "idiopathic neuropathies" can now be explained by genetic mutations. Finding these genes will help us formulate treatments in the future. The scientific correlation between a genetic mutation and its clinical manifestations is fascinating to me. There is a lot to achieve in scientific knowledge and

treatments in those with CMT. Also, CMT involves the most wonderful, kind and intelligent people I have ever met. I enjoy helping and treating patients and their families who have CMT.

Q: TELL US WHAT PATIENTS WILL EXPERIENCE WHEN THEY COME TO THE HNF CENTER OF EXCELLENCE (COE) AT THE ADVENTHEALTH NEUROSCIENCE INSTITUTE?

The AdventHealth Neuroscience Institute is fully committed to patient excellence, care, and exceeding expectations. Patients will experience a team that is superb beyond expectations. If they are coming from far, they will be catered to with our destination team. When they arrive, they will experience an environment full of love, kindness, friendliness, and softness. They will observe an amazing facility, receive an in-depth physician evaluation, electrodiagnostic testing, genetic testing, and opportunities to get all the multidisciplinary therapies that they need. They will also be involved in any research studies that we may have.

Q: WHAT ARE THE BENEFITS OF A PATIENT BEING EVALUATED AT A HNF COE?

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

Q: WHAT OTHER DOCTORS WILL PATIENTS BE ABLE TO SEE WHEN THEY VISIT THE COE?

Pain physician, podiatrist, orthopedic surgeon, social worker, physical therapist, occupational therapist,

spiritual counselor, research evaluation, pulmonologist, orthotist, and much more as needed.

Q: WHAT KIND OF SUCCESS STORIES HAVE YOU SEEN SO FAR WITH CMT PATIENTS?

There are many success stories. Some stories involve finding rare genetic mutations for patients and their families, which is important for their future generations. Other success stories involve having patients actually do certain therapies such as physical therapy and occupational therapy, which can help improve leg strength, gait, balance, and hand strength.

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

Q: HOW CAN PATIENTS MAKE AN APPOINTMENT AT THE COE?

Patients can call our clinic at 407-303-6729 and specifically request for a visit with Dr. Jerath in the CMT clinic. Multiple family members including children are welcome. Patients can also email Dr. Jerath at Orl.neuromuscular.medicine@adventhealth.com if they need to communicate with her for any reason.

HNF's CMT Centers of Excellence

The national network of HNF-designated Centers of Excellence (COE) provides patients with resources to find hubs of expertise in caring for and treating CMT, as well as locations where CMT research is being conducted. Our primary goal is to ensure care results in positive outcomes for each individual patient's clinical experience. We are honored to partner with these premier Centers and their leading experts to improve the future for people with inherited neuropathies.

CALIFORNIA

Cedars-Sinai Medical Center
Los Angeles, CA
Contact: Dana Fine
310-423-8497 Dana.Fine@cshs.org

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

CONNECTICUT

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

FLORIDA

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

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

University of Miami
Miami, FL
Contact: Meri Jaime (for appointments)
305-243-7400 MJaime@med.miami.edu

ILLINOIS

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

KANSAS

University of Kansas Medical Center
Kansas City, KS
Contact: Nicole Jenci
913-945-9934 njenci@kumc.edu

MASSACHUSETTS

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

MICHIGAN

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

MINNESOTA

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

MISSOURI

St. Louis University Medical Center
St. Louis, MO
Contact: Susan Eller
314-977-4867 ellersc@slu.edu

MU Health Care:
Columbia, MO
Contact: Dr. Raghav Govindarajan
573-882-1515 govindarajanr@health.missouri.edu

NEW JERSEY

Hackensack University Medical Center
Hackensack, NJ
Contact: Annerys Santos
551-996-8100
Annerys.Santos@HackensackMeridian.org

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

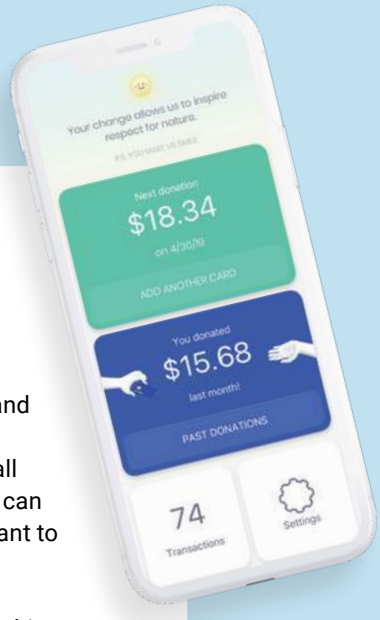
NEW YORK

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

WASHINGTON

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

* Pediatric Center of Excellence



Help Support HNF with the RoundUp app!

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

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

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

Thanks,
Courtney

HEREDITARY NEUROPATHY FOUNDATION

401 Park Avenue South
10th Floor
New York, NY 10016
hnf-cure.org

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Events

11th Annual Card Party for CMT2A

On February 7, 2020, H.E.L.P. (Help Elliot Live Proud) Fund supporters participated in the 11th Annual Card Party Brunch and Boutique at a new venue, the Boca West Country Club, in Boca Raton, FL. Record numbers flocked to the pristine country club for a day filled with gourmet food, a boutique, raffles, cards and an amazing silent auction. HNF longtime supporter, Iris Adler, is dedicated to making this event a success year after year, and we are grateful for her dedication. Many new faces attended this year, and new faces equal spreading awareness for Charcot-Marie-Tooth. One regular attendee noted, "One of my favorite charity events year after year — and what a great cause." HNF is proud to report that the event raised more than $30,000 to fund research for CMT2. The Adler family would like to thank the participants, volunteers and businesses who donated and those who were unable to attend, but contributed. It's thanks to people like you that we WILL find a cure for CMT!

For more information on the H.E.L.P. Fund or to support CMT2 research: www.hnf-cure.org/h-e-l-p-for-cmt/

Get Involved and Join us at an Upcoming HNF Event

Movement is Medicine™ Summit Orlando

**** POSTPONED ****

Join us for a day of CMT-friendly exercise classes, workshops, and one-on-one consultations. Lunch will be provided.
Register today: www.move4cmt.org

Euchre Tournament, Rochester, NY

**** POSTPONED ****

Join us for a fun afternoon of Euchre with cash prizes, refreshments & more! All to benefit CMT6.
Register today: www.curecmt.org

Jaxon's Crusaders Clay Shoot, Fort Worth, TX

**** POSTPONED *** *New Date June 20, 2020*

All ages are welcome to attend this family-friendly event. We will have live music, face-painting, and plenty of kid-friendly activities. All proceeds benefit Leigh's syndrome gene therapy research for Jaxson Flynt.
Register today: JaxClayShoot.givesmart.com

PA 4 CMT, Newtown Square, PA

**** POSTPONED *** *New date September 18, 2020*

The evening will be filled with cocktails, food, music and amazing silent auction items. Take a chance on the 50/50 raffle!
Join us: PA4CMT.givesmart.com

Team CMT Spin, New York City

May 2, 2020

Spin with CMT Warrior & Instructor, Marc Daigle, to support CMT research!
Register today: https://bit.ly/3aGaA87

TD Bank Five Boro Bike Tour, New York City

May 3, 2020

HNF Riders get VIP treatment, which includes breakfast and lunch, as well as priority start and special bike parking.

Million Dollar Bike Ride, Philadelphia, PA

June 13, 2020

Register to ride with Team CMT on June 13th. Riders can select a 13, 34 or 72 mile route and funds raised will be matched by the UPenn Medicine Orphan Disease Center (up to $30k)!
Register today: https://bit.ly/2W1LvAA

Movement is Medicine™ Summit, Phoenix, AZ

November 13-14, 2020

Join our Summit waiting list for 2020 – space is limited! Sign up to receive notifications about this event.
Learn more: https://bit.ly/2TBFOaJ

PHARNEXT

Publication of this newsletter was made possible with the financial support of Pharnext.