HEREDITARY NEUROPATHY FOUNDATION

FALL 2020

cmtupdate

CAPTURING CMT WITH NEW TECHNOLOGY

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New Developments: CMT4 and CMT6
Gene Therapy
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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 2020

Allison T. Moore
Founder and CEO
Hereditary Neuropathy Foundation

I hope this message finds you well and your family safe and healthy. In these unprecedented and tumultuous times, it is more important than ever to rally around our families and loved ones as we head into the holidays.

Our HNF community members continue to inspire me with their resilience and ability to adapt to the fluid situation that COVID-19 presents to us. As people living with CMT and care partners, we know all too well about the challenges and obstacles that we must overcome on a daily basis just to live our lives. Our HNF Board of Directors, Therapeutic Research In Accelerated Discovery (TRIAD) Council and staff are committed to supporting our community with innovative programs that help us not only cope with our condition, but thrive...we will not slow down!

If anything, HNF has grown more this year than any year previous. In 2020, we have diligently explored both the challenges and opportunities of new drug discovery, examined the barriers in patient care, and found new ways to raise funds for important research. In this issue of the CMT Update, you will read all about our continued commitment to moving the needle towards treatments and how best to support research, clinical trials, and improved patient care.

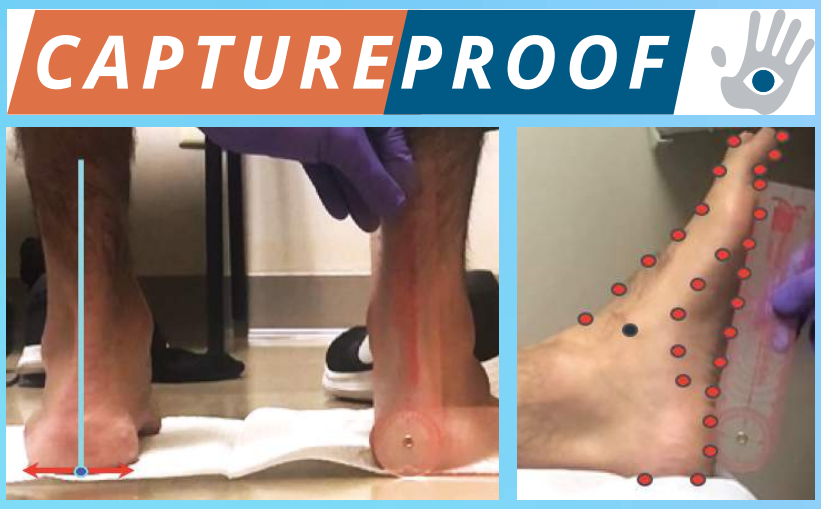
I am thrilled to announce that HNF has launched an innovative natural history study that will be a game-changer in advancing treatments for CMT. The CMT foot is typically characterized by the pes cavus (high arch). HNF is moving forward with a state of the art virtual clinical pilot study to capture measurable images of your feet with an iPhone and the CaptureProof app that includes a Smart Medical Camera™. Our pilot study will collect other important data on foot structure, gait, hand function and more. The data will help inform a better understanding of CMT characteristics, symptoms and function to support better clinical trial design. We strongly believe in giving back to our community when participating in research. With that in mind, you'll have the option to have your data shared with your treating medical professional and/or have the opportunity to set up virtual telemedicine appointments with one of our leading CMT experts. This important study has the potential to improve standard of care and make a meaningful difference to the countless people living with CMT.

Please read more about this in our Featured Article and consider funding our innovative approach to accelerate the path to a cure.

With your support, we will get there faster!

With gratitude,

Allison T. Moore



IF A PICTURE IS WORTH A THOUSAND WORDS, **THEN A VIDEO IS WORTH A MILLION**

We may all have that secret. Our parents probably first noticed it when we were very young. We typically hide it, but when we are in private, social media CMT groups it slowly comes out: "Show me your feet". "What do you do about shoes for your high school prom or your wedding?" "My son is a toe walker." "What shoes work best with AFOs?" It's our feet! We all have abnormal looking feet. In a published study, the probability of a patient with bilateral cavovarus feet being diagnosed with CMT disease, regardless of family history, was 78%. A family history of CMT disease increased the probability to 91%.[1] With all the varying symptoms between types of CMT, and even within the same type of CMT, an overwhelming majority of us have foot deformities and our feet were the first sign that something was wrong. Foot deformities lead to an abnormal gait, drop foot, high steppage gait, loss of balance, ankle weakness and sprains. As the disease progresses, foot deformities lead to a lifetime of physical and emotional pain.

It's time we stop this disease in its tracks!

In 2016, HNF ran an ad in Neurology Reviews, a peer review journal targeted at Neurologists, Primary Care Physicians, Nurse Practitioners/Physician Assistants, Genetic Counselors and Geneticists with a photo of a "CMT foot" and text that read, "Why does it take the average patient 2 years to receive a diagnosis?" Dr. Amro Stino, a leading CMT expert neurologist familiar with the many research papers about the CMT foot, asked his colleague, Dr. Said Atway, also a leading CMT expert and podiatrist, if there might be a way to define the "CMT foot" with measurements. Together, the two doctors (along with several others) developed and launched the research study and published their interesting results.[2] As you can imagine, they had trouble finding enough CMT patients to make an impact.

[1] Prevalence of Charcot-Marie-Tooth Disease in Patients Who Have Bilateral Cavovarus Feet Mary K. Nagai, MD, PhD, Gilbert Chan, MD, James T. Guille, MD, S. Jay Kumar, MD, Mena Scavina, DO, and William G. Mackenzie, MD

While HNF was designing the Movement is Medicine™ Summit and Arizona State University (ASU) CMT gait study, we were introduced to Meghan Conroy and her company, CaptureProof – the visual health record for HIPPA compliant photo and video communication between patients and doctors. Together, we were looking for signs and symptoms to capture, and we were reminded of Drs. Stino and Atway's published paper and that's where it all began! Both researchers have now joined our team and are working with HNF and CaptureProof with the goal of expanding their study of the pes cavus CMT foot. The basis of the study is to capture important visual data, very simply with an iPhone! We're calling it the "Foot Calculator".

A picture is worth a thousand words…especially if it's taken correctly. HNF, in partnership with CaptureProof, is bringing innovative and powerful computer learning capabilities to give you the power to capture and document your CMT story to improve the health care you receive. CaptureProof uses advanced computer vision in the live camera to give instant feedback on quality of photos and videos. With the CaptureProof App, patients will be directed to use the Foot Calculator to capture specific photos and videos with the Smart Medical Camera™ to identify and track progression of pes cavus and the CMT foot. Many CMT signs and symptoms will be captured in photos and videos over time to synchronize with telemedicine and/or in-office visits. Not only is this a safer way to visit with your doctor, but we know that one of the most frustrating things about CMT is being able to find a doctor that understands how to treat it. With telemedicine it's becoming much easier. Essentially, you'll be putting a CMT Center of Excellence in your living room!

This project will have an important impact on the diagnostic journey, clinical trials (cost-effectively tracking progress of more patients without boundaries) and access for underserved populations. It will also serve as a Natural History Study to track the degenerative progression of the disease. The app will also include measurements such as, gait analysis videos, finger dexterity, tremors and more.

Join GRIN today to be considered for the pilot study.
www.neuropathyreg.org

If you value this work being done by HNF, please make a donation to our research programs TODAY.

DONATE: CLICK HERE

[2] Foot Measures in Patients with Pes Cavus with and without Charcot–Marie–Tooth Disease: A Pilot Study Amro M. Stino, MD, 1 Said Atway, MD, 2 Michael Anthony, MD, 2 David Kline, PhD, 3 And John T. Kissel, MD, PhD 11 Ohio State University Wexner Medical Center Department of Neurology, Division of Neuromuscular Medicine, Columbus, Ohio, USA 2 Ohio State University Wexner Medical Center Department of Podiatry, Columbus, Ohio, USA 3 Ohio State College of Medicine Department of Biomedical Informatics Center for Biostatistics, Columbus, Ohio, USA Accepted 17 July 2018

Principal Investigators Spotlight:
Dr. Amro Stino and Dr. Said Atway

Amro Stino, MD is an Assistant Professor in the Department of Neurology in the University of Michigan Medical School, Division of Neuromuscular Medicine. Dr. Stino has expertise in the treatment of neuromuscular disorders, with special clinical and research interest in the care of patients with acquired and hereditary peripheral neuropathy. He served as site investigator in the Pharnext PXT3003 trial in CMT1A. He serves as site principal investigator on multi-center studies and is leading an investigator initiated phase 1b study as well. Dr. Stino is an advocate for patient awareness and education in CMT and peripheral neuropathy through patient advocacy organizations.

Said Atway, DPM received his Bachelors of Science Biology from Youngstown State University. He received his medical degree from Ohio College of Podiatric Medicine. Following medical school Dr. Atway completed his PSR-36 residency program at The Ohio State University Wexner Medical Center.

Following his residency, Dr. Atway joined The Ohio State University College of Medicine in 2009. He currently serves as Co-Director of Limb Preservation, he is a Researcher for the Center for Regenerative Medicine and Cell-Based Therapies and serves on the Wound Care Panel for the Comprehensive Wound Center. Dr. Atway is an Associate Professor for the Department of Orthopaedics.

Dr. Atway specializes in Flatfoot, Plantar Fasciitis, Foot Ulcer, Foot Injuries, Foot Deformities, Diabetic Foot, Hallux Valgus, Orthopaedics, Limb Preservation and Surgical procedures. He also focuses on foot impairments as a result of peripheral neuropathies and early detection and diagnosis of Charcot-Marie-Tooth (CMT). His expertise in measurements defining the Charcot-Marie-Tooth (CMT) pes cavus arch was crucial in the development of the Foot Calculator.

Dr. Atway has authored and co-authored 15 peer-reviewed articles in scientific journals, book chapters, and scientific presentations. He has presented at multiple regional and national society conferences. He is a current member of the American College of Foot and Ankle Surgeons, Ohio Foot and Ankle Medical Association, and the American Podiatric Medical Association. He serves on the Exam Preparation Task Force and is an Ad-hoc Contributor for the American Board of Foot and Ankle Surgeons. Dr. Atway is also a reviewer for Clinical Research on Foot and Ankle and for the International Wound Journal.

TRIAD
THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY

HNF Newly Formed **TRIAD Council**

The Therapeutic Research In Accelerated Discovery (TRIAD) Council is composed of CMT thought leaders engaged in collaborative planning and decision making to provide regular guidance and direction to HNF's research strategy. This network of professionals, led by Dr. Lucia Notterpek, HNF's Chief Scientific Officer, meets quarterly. They will review grant proposals, provide expert guidance and assess project outcomes to advance therapeutic development for CMT.

This diverse group of experts was specifically chosen to represent the many facets of CMT research, treatment and drug development. We sought to recruit researchers who are studying CMT, but also researchers who are studying similar, related diseases and co-morbidities. We sought out clinicians who treat CMT in addition to other similar diseases. Also included were key opinion leaders in outcomes research, gene therapy and the pharmaceutical industry.

READ MORE ABOUT EACH COUNCIL MEMBER:
Click Here!

It starts in infancy and lays dormant in the body until one day... the symptoms start to appear.

Everything seems okay until your...

Even if you are lucky enough to get a proper diagnosis of this genetic disease—Charcot-Marie-Tooth—there is no cure, and it is progressive.

But the Hereditary Neuropathy Foundation is on track to finding treatments and cures! **LEARN MORE AT** **www.hnf-cure.org**

New Board Member Dominic Hadeed

Mr. Dominic Hadeed currently leads or has investments in a diverse group of successful enterprises in the Caribbean including Blue Waters Products Limited, Blue Waters St. Lucia Limited, Fabric Land Limited, Domhad Properties Limited and shareholding investments in other corporate companies. Mr. Hadeed also holds membership in professional bodies such as the Young Presidents Organization and remains one of the few members from Trinidad and Tobago in this global community of CEO's under the age of 45, with approximately 22,000 members in over 125 countries.

In recognition of his outstanding achievements, in 2015 Mr. Hadeed was awarded 'The Ernst and Young's Master Entrepreneur of the Year Award' and became EY's country winner for Trinidad and Tobago. In 2016, Hadeed represented Trinidad and Tobago at 'The World Entrepreneur of the Year Award' gala event which was created by EY to recognize the accomplishments of entrepreneurs around the globe. Under his leadership, Blue Waters Products Limited emerged winner as the "*Best Managed Company 2019*" by Deloitte & Touche. Dominic has CMT2A and resides with his family in Trinidad and Tobago.

$100,000 Match for CMT2A

HNF has received a $100,000 pledge as a match challenge to help accelerate research for CMT. The gift is earmarked for CMT2A which is the second most common type of CMT. These funds will continue to advance the development of selective histone deacetylase (HDAC) inhibitors for enhanced therapeutic outcomes. HNF has supported this approach for almost a decade. We are getting closer, but we still need your help! It usually takes up to 14 years and millions of dollars to bring a drug to market. Our efforts are finally paying off.

TIMELINE:

- **2010:** Developed a CMT2A Mouse Model
- **2017:** Partnered with University of Sheffield and Acetylon Pharma to conduct testing in CMT2A Zebrafish
- **2019:** Partnered with StarWise Biotech and the University of New Zealand to optimize Vital Drug Experiments
- **2020:** Published findings in the *Experimental Neurology: HDAC6 inhibition promotes α-tubulin acetylation and ameliorates CMT2A peripheral neuropathy in mice*
- **2020:** IP assigned from StarWise to undisclosed biotech Company, to optimize RX.

We are eager to hit the $100,000 goal by year end to keep up the momentum. Please consider donating during this crucial time as we advance this potential therapy to patients. **Every dollar raised up to $100,000 will be matched, giving us a total of $200,000 in funding!**

A special thank you goes out to our newest board member Dominick Hadeed for his generous pledge in the amount of $100,000. We cannot thank him enough for joining our team. We also want to highlight two other special families that continue to tirelessly raise funds for CMT2A.

TO LEARN MORE VISIT:

The Adler Family:
Help Elliot Live Proud (H.E.L.P.)

The Caldarone and Sidoti Family:
Grace's Courage Crusade (GCC)

HNF works for you, the patients and we will not stop until we deliver treatments. We care deeply about our community and donors and have made every one of your dollars count to get us here. There has never been a more exciting time for CMT2A research. A treatment is within our reach. **Let's do this! Donate today.**

Click here to donate to CMT2A

Email courtney@hnf-cure.org to take the first step to customize your very own family page to support your type of CMT. The HNF team will guide you and provide everything you need.

If you are interested in joining the HNF board please email allison@hnf-cure.org

TRIAD
THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY

Gene Therapy Program:
New Developments with CMT4A and CMT6 – HNF Collaborates with Leading CMT Scientists

HNF has awarded leading induced Pluripotent Stem Cells (iPSC) scientist, Dr. Mario Saporta at University of Miami (an HNF Center of Excellence) a $160,000 grant to support gene therapy studies of CMT4A (GDAP1) and CMT6 (c12orf65). With demonstrated positive results, HNF and top CMT researchers are moving the needle towards a cure. These innovative projects will play an important part in HNF's Therapeutic Research In Accelerated Discovery (TRIAD) research strategy of translating viral vector gene replacement to treat two types of CMT.

Dr. Saporta's Stem Cell Work: Dr. Saporta's lab specializes in utilizing human CMT patient skin samples and puts them through a stem cell reprogramming process to create neurons (nerve cells). He is developing motor neuron assays to better represent the human biology of both CMT4A and CMT6. These models will help in testing specific gene therapies for each of these sub-types of CMT. Both CMT4A and CMT6 are axonal recessive types of CMT, which are associated with the complete loss of function of the GDAP1 and C12orf65 genes, respectively. Therefore, these CMT subtypes are ideal conditions for gene replacement therapy, where a missing gene is put back into the body using viral carriers.

What is CMT4A?

A rare subtype of CMT caused by recessive mutations in GDAP1. Patients with CMT4A usually present CMT symptoms early in life and tend to have a more severe functional disability.

What is CMT6?

A rare subtype of CMT caused by recessive mutations in the C12orf65 gene. Mutations in C12orf65 can present in multiple ways, depending on several biological factors, including central nervous system symptoms (encephalomyelopathy or Leigh Syndrome), early onset optic atrophy, peripheral neuropathy, and spastic paraparesis. CMT6 is a progressive and very severe form of CMT, often presenting with autonomic distress, blindness and in some cases, early death.

Animal Models to Validate Gene Replacement

HNF Develops Animal Models to Further Validate Gene Replacement Strategy: The dedicated research team of Robert Burgess, PhD, Steven Gray, PhD, Dianna Willis, PhD and others will be instrumental from development to human trials. Each researcher plays a vital role in successfully translating this complex process into a living model.

CMT6 Models: In 2017, HNF partnered with Dr. Burgess at Jackson Labs and Dr. Gray at UNC, (he now is at UT Southwestern) to develop a gene therapy approach to cure C12orf65.

Dr. Burgess worked on the development of a mouse model of C12orf65, but the pups would die once the mouse developed C12orf65. Dr. Burgess quickly re-evaluated the path and engineered a cell model to replicate C12orf65 as an assay to establish proof of concept that gene therapy could be a viable path for a cure. This assay is now at Dr. Saporta's lab, along with the viral vector that Dr. Gray developed. Next steps will involve testing the vector for potential rescue of mitochondria malfunction – the energy factory of our cells, which causes severe autonomic and motor dysfunction in patients with this disease.

CMT4A Models: HNF has successfully developed a rat model in collaboration with Envigo, a leader in producing customized animal models using CRISPR-edited technology. The novel rat model is being characterized by Dr. Willis at Burke Institute in New York. She is an expert in investigations of axonal biology, the long processes that are key for communication between nerves and muscle cells, and will lead the studies. In addition, we plan to distribute the preclinical model to enable increased access to be used as a research tool to accelerate GDAP1 Research.

HNF Launches Natural History Studies: Patient Engagement is key to understanding the natural history of their experience from the time of diagnosis through present day. To better understand HNF partnered with CaptureProof, a digital platform using photos and videos for communication between patients and researchers. This collaboration will help to document a patient's CMT symptoms and functionality for the development of trial protocols.

Thank you to all our GDAP1 & CMT6 families for your participation and support thus far. We are off to a good start and couldn't have gotten here without the support of our donors. Together we are stronger and a part of history in the making. We know firsthand as patients ourselves, the daily challenges of living with CMT and how overwhelming this journey can seem, but together, we can do this! Please know that we are here to support and guide our families each step of the way. So please consider making a donation and/or building a campaign targeting families and friends to support our research.

To Donate: CMT 4 (GDAP1) Click Here!

To Donate: CMT6 (C12orf65) Click Here!

Act of Kindness: From Strangers to Instant Friends

On October 14, 2020, Winter School athletes in Wisconsin hit the Tuscobia Trail to raise much needed funding for HNF's gene therapy program in support of Alana Kohler. This amazing school reached out to HNF after they saw a media piece on Alana Kohler who has GDAP1 (CMT4). They were responding to the family's quest to raise funds for HNF's gene therapy program.

The Kohler family drove 150 miles for the event and handed out water and Alana's home-baked goodies at two water stations along the trail. The event raised over $10,000 through a gift basket raffle and an 11-mile run/walk on the Tuscobia Trail from Winter to Radisson for a girl they didn't know with a disease they'd never heard of.

The event give Winter Student athletes something to look forward to and served to redirect their focus from COVID-19, distance learning, and canceled daily practices and competitions.

Thank you to Winter School's coaches, student athletes and the Winter School community for your dedication to finding a cure for Charcot-Marie-Tooth disease and for your kindness to the Kohler Family.

Learn more about GDAP1 patient Alana Kohler's story in community section on page 18.

DONATE TO HNF'S GENE THERAPY PROGRAM FOR GDAP1 (CMT4):
Click Here

TRIAD
THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY

Allison Moore & Estela Lugo

GDAP1 Hits Close To Home

BY ALLISON MOORE

Over ten years ago I met the most incredible young woman, Estela Lugo. She is now 40 years old, a mother of two children and she lives with CMT, just like me. Luckily, Estela lived close by where I grew up, and we bonded instantly. We became fast friends and later, Estela joined the HNF team.

Unlike me, Estela grew up knowing at an early age that she was different. At age 3, Estela was diagnosed with an axonal form of CMT. Mine is demyelinating. Although they often present similarly, knowing the mutation is important when thinking about a potential treatment. Each CMT gene is, from a mechanistic standpoint, a really different disease. I explained to Estela, who I wanted so badly to cure, that she should consider genetic testing to find her sub-type of CMT. Without knowing her affected gene, she would not be eligible for upcoming clinical trials. It was back in the 90's when she first did genetic testing and the results were inconclusive. She was told she had CMT type 2. After nudging her year after year, she finally gave in! Estela was officially diagnosed with a mutation in GDAP1, a recessive type of CMT, called CMT4A.

Now that we have a definitive diagnosis, we are excited to have her included with the rest of our growing GDAP1 families. For more information on Genetic Testing and CMT, watch our exclusive webinar with Athena Diagnostics.

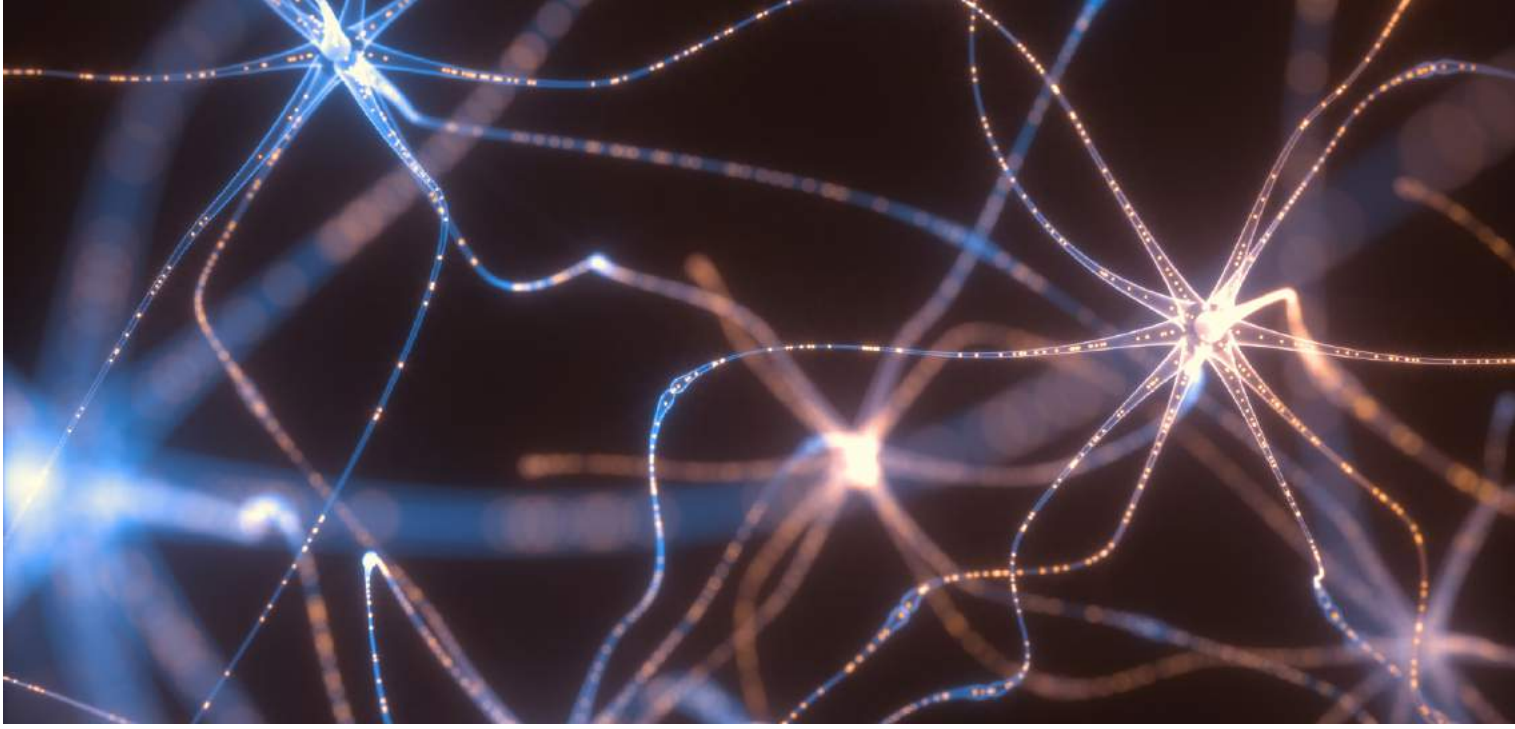
If you or a family member have CMT4A (GDAP1), reach out to the HNF Team and learn more about how you can help with this project! Email estela@hnf-cure.org

DONATE TO GDAP1: Click here!

Mouse Models of Human CNTNAP1 Mutations: Strategies to Rescue and Restore Functions

MANZOOR A. BHAT, M.S., PH.D., DEPARTMENT OF CELLULAR AND INTEGRATIVE PHYSIOLOGY
UT HEALTH SAN ANTONIO, TX

Myelination of nerves has the sole purpose of allowing axons to propagate nerve impulses over long distances in a saltatory manner. Peripheral or central neuropathies or diseases that affect nerve conduction are the leading cause of non-injury related neurological and neuromuscular disability and affect over 2.5 million people worldwide. In healthy individuals, the voltage-gated ion channels become enriched at distinct regions along myelinated axons, allowing them to drive rapid propagation of nerve signals for cognitive functions as well as motor and muscle movements. Any perturbations in the structure of the myelinated nerves that reduce nerve conduction result in devastating disorders leading to loss of nerve and muscle functions. Currently, there are no therapeutic strategies available to restore myelinated axon structure and function. Moreover, there is also a lack of understanding of the critical therapeutic timeframe within which restoration of axonal functions must occur to recover nerve conduction, and ultimately regain lost mobility. With the decline in nerve function, the target muscles eventually begin to atrophy forcing the subjects to become wheelchair bound for the rest of their lives, thus impacting their quality of life and



putting emotional and economic burden on the patients and their families.

Work from our group and others over the past two decades has identified key proteins that are central to the organization of the myelinated axons into functionally distinct domains defined by the presence of specific protein complexes that are essential for rapid nerve action potential propagation. One of these regions is named the "paranodal domain" which contains the axo-glial junctions (AGJs) that are established by Contactin-associated protein 1 (Cntnap1) and Contactin from the axonal side and Neurofascin 155 from the myelin side. Mouse models that have been genetically manipulated to become deficient in Cntnap1 or Contactin or Neurofascin 155 all become paralyzed with severely reduced nerve conduction leading to muscle atrophy, and die within 3-4 weeks after birth. These mouse mutants also show loss of the paranodal domain and disorganization of the myelinated axon structure and function.

Recently, mutations in human Cntnap1 gene were identified in a number of children that are associated with severe myelin defects further highlighting the importance of the Cntnap1 protein in nerve structure and function. Most of the children with Cntnap1 mutations display a broad spectrum of disabilities ranging from extreme muscle weakness to mild weakness, and all show developmental delays. Some children may develop typical phenotypes of the Charcot-Marie-Tooth disease with symptoms of the peripheral neuropathy. Given the broad range of mutations found across the Cntnap1 gene, it is clear that all these mutations alter Cntnap1 protein function leading to the disorganization of the paranodal domain and the myelinated axon structure. Unfortunately, there are no treatments or therapeutic strategies available to help the afflicted children to regain nerve functions and to restore mobility to live an everyday normal life. Genomic DNA analyses of the families have revealed that in most cases one parent carries a mutation that leads to a complete loss of one of the Cntnap1 alleles, and another parent carries a mutation that causes a single amino acid change in the Cntnap1 protein. The children thus only have the Cntnap1 protein with the amino acid change that affects the Cntnap1 protein function in the nerve fibers. To begin to address how the children with Cntnap1 mutations could be helped to reduce their disease burden in the near future, we have generated mouse models with exactly the same mutations as some of the children have in their Cntnap1 gene. We have created the same mouse mutant combinations in which one allele of the mouse gene is absent and the other allele has the child's mutation. These mice are currently being analyzed for defects in nerve conduction and nerve fiber structure to conclusively establish the impact of these single amino acid changes in Cntnap1 on its function and the physiological properties of the nerve fibers. Each mouse line is independently analyzed using a battery of phenotypic methodologies. We have also generated genetically modified mice that will allow us to produce normal Cntnap1 protein in the mutant mice that carry human mutations to establish whether this Cntnap1 protein can rescue the mutant animals at various times after birth and restore nerve function. To further establish a gene delivery method that could produce the normal Cntnap1 protein in the mutant animals, we have generated Lentiviral vectors that carry the full-length Cntnap1 gene. The recombinant viruses will be injected into the mutant mice to determine the efficacy of Cntnap1 gene delivery by viral vectors and the restoration of lost functions. Collectively, the mouse models of human Cntnap1 mutations offer the best hope to test restoration strategies as "proof of principle" towards human applications in the treatment or management of Cntnap1 mutations to restore nerve functions.

Acknowledgments
The work on Cntnap1 mutations and developing therapeutic strategies has been sponsored by the families through the Hereditary Neuropathy Foundation. This support is vital for testing restoration strategies to these devasting human conditions.

DONATE TO CNTNAP1: Click Here!

TRIAD
THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY

Free CMT Genetic Testing for a Common Axonal type of CMT to Prepare for Clinical Trial

A new type of Charcot-Marie-Tooth (CMT) has recently been identified caused by mutations in the SORD gene. This is likely the most common recessive type of CMT; and the best part is that drugs are likely available that have been developed, in principle, for diabetic peripheral neuropathy.

Under the leadership of Dr. Stephan Züchner, a world renowned geneticist and neurologist at the University of Miami, one of HNFs Center of Excellence along with his team, including Drs. Andrea Cortese, Grace Zhai, and Adriana Rebelo have made the groundbreaking discovery. This study was substantially supported by the Inherited Neuropathies Consortium led by Dr. Michael Shy at the University of Iowa. He and other Researchers think that at least 3,000 – 5,000 people in the United States—and more than 60,000 worldwide—have this type of CMT.

Dr. Züchner's research has shown that drugs approved for other diseases that target this SORD gene may show promise as a viable treatment for CMT. If so, it would be possible to accelerate the clinical trial timeline and skip straight to Phase 2 since the drugs have already gone through Phase 1 testing for other diseases and indications.

HNF is also supporting the development of a SORD rat model for testing the new drugs for additional validation studies prior to human trials. These efforts are well under way and we will keep you posted as the project develops.

"We are so excited for Dr. Züchner's discovery and the potential of a therapy that we are supporting the collaboration by funding the development of a SORD rat model to support more research to validate these drugs prior to human trials."

– Allison Moore, CEO of HNF

"This remarkable discovery has been possible because of CMT patients participating in our genetic studies, " said Dr. Züchner. "For the next phase we are committed to offering free SORD gene testing to screen undiagnosed CMT patients in the hopes of more patients receiving a confirmed CMT diagnosis as well as identifying more patients in preparation of clinical trials."

If you are interested in participating, please reach out at allison@hnf-cure.org and join the patient registry, Global Registry for Inherited Neuropathies (GRIN). The criteria for screening are:

1) you have received a diagnosis of CMT, but genetic testing did not reveal with certainty the responsible gene;
2) your parents never had CMT symptoms and
3) you may have a sibling also affected with CMT symptoms.

TO SUPPORT AND ACCELERATE A PATH TO TREATMENT, DONATE:

Click Here!

GLOBAL REGISTRY FOR INHERITED NEUROPATHIES

3 Reasons to Join the Global Registry for Inherited Neuropathies – GRIN!

Started in 2013 in partnership with Hannah's Hope Fund, the Global Registry for Inherited Neuropathies - GRIN - has been collecting invaluable, self-reported natural history data directly from CMT patients and/or their caregivers.

This data has helped HNF and its partners in industry, academia and government identify previously unknown genotype/phenotype correlations, uncover important comorbidities such as pain or respiratory issues, and allowed us to target our research spending based on actual patient need.

By participating in GRIN, you are joining a community of patients dedicated to helping us find treatments and cures for CMT and other inherited neuropathies.

Just some of the reasons to join are:

1

Joining GRIN can help you get fast-tracked into important clinical trials and studies, including HNFs pilot study collecting important visual data to enhance diagnosis and treatment, as well as support clinical trial design.

2

You are empowering our global network of researchers with important self-reported data that can be used to identify potential new avenues for therapeutic development.

3

Your participation benefits the CMT community as a whole - we all win together!

Help us in our fight to find a cure for CMT and other inherited neuropathies – join GRIN today!

Click here!

NEUROLOGY REVIEWS

Meet Elizabeth Katz
Publisher, *Neurology Reviews*

How does HNF educate Healthcare Providers? Why is this so important?

HNF does a great job of educating our readers – Neurologists, Primary Care Physicians, Nurse Practitioners/Physician Assistants, Genetic Counselors and Geneticists - about CMT.

Even though CMT is one of the most common rare diseases, it is often misdiagnosed or under diagnosed. Part of the problem is that the first symptoms of CMT (foot drop and a high-stepped gait, curled toes, frequent tripping or falling, or loss of muscle) can often be misdiagnosed as another disorder, including Multiple Sclerosis (MS) (Dr. Charcot also identified MS as well), or other peripheral neuropathies.

It is important that neurologists are made aware of the various signs and symptoms of CMT, even though they may seem insignificant (e.g., a child having trouble tying her shoelaces, frequent tripping or problems with balance). HNF created a Continuing Medical Education (CME) course for this purpose.

In addition, HNF works closely with *Neurology Reviews* and our annual *Rare Neurological Disease Special Report*, to provide neurologists and other healthcare providers with a wealth of valuable resources, the latest clinical research, announcements of clinical trials, and scientific information all with the goal of shortening the time to diagnosis and improving the quality of life for patients and their family members dealing with CMT. Through the surveys conducted by HNF, it was discovered that 65% of patients with CMT suffer from pain. This was important information that was distributed via *Neurology Reviews*.

Over the last 6 years *Neurology Reviews* has conducted a Readership Survey post-publication of our annual *Rare Neurological Disease Special Report* that is developed in collaboration with NORD, in which HNF is also an advertiser. These Readership Surveys help us to understand what articles our readers are engaged in, how they want to learn about rare diseases, where they go for information about rare diseases, and what rare diseases they want to learn more about. CMT has consistently been ranked as one of the top rare diseases they are most interested in and want to learn more about.

In addition, in the April 2020 Readership Survey, we also included an Addendum Question developed by HNF to be included in the Readership Survey and sent to neurologists, medical geneticists and genetic counselors. See the summary of results, below.

HNF provides a wonderful and unique feeling of community, inspiration and advice on how to improve quality of life, physical fitness and wellness, and the latest research and support for the patients, their families and their caregivers. Our neurologists, and other healthcare providers treating these patients, can then share with their colleagues and their patients.

Affordable genetic testing options and complementary therapies (i.e. exercise protocols) would be most useful to respondents' Charcot-Marie-Tooth patients.

Resources for CMT Patients	(Total Responding = 99)
Affordable genetic testing options	57%
Complementary therapies (i.e. exercise protocols)	43%
A list of Centers of Excellence	34%
Early access ("Compassionate Use") to drugs	33%
Enrollment in patient registries (i.e. Global Registry for Inherited Neuropathies (GRIN))	32%
Referral resources for other treating HCPs (i.e. PTs, orthopedic surgeon, etc.)	28%
I do not treat Charcot-Marie-Tooth (CMT) patients	15%
None of these	1%
Other (please specify)	0%

QUESTIONS? Click Here!

HNF Partner Launches New Logo

The Inspire CMT Online Support Community connects patients, families, friends and care partners for support and inspiration. HNF sponsors the community and is an Inspire Trusted Partner. Inspire has recently freshened up its logo and we think its fantastic!

Inspire's mission is to accelerate life-changing discoveries through a vital community of connected patients. They believe in genuine connections, an atmosphere of trust, a desire for wellbeing, and meaningful impact. Inspire is the leading social network for health that connects patients and care partners in a safe, permission-based manner.

Finding other people with the same diagnosis can be difficult, especially for people with rare diseases like CMT. We hope that you'll take the time to join the Inspire CMT Support Community and connect with 8,000 others who share our concerns related to diagnosis, managing symptoms, and treatment options such as bracing, orthotics, and exercise. Most of all, you will find comfort that you're not alone.

It's easy to get started. After you create a personal profile, you can immediately start participating in discussions. Extensive privacy features allow members to control exactly how their personal information is shared with others.

Join CMT Inspire Online Support Community:
https://www.hnf-cure.org/online-support-community

CMT-Connect Pain Webinar

It's no secret that PAIN can play a major role in the lives of many CMT patients. Despite its prevalence there is still little guidance on how families can identify and address the wide spectrum of pain our community continues to experience.

HNF aims to help close this gap with a new **CMT-Connect Webinar Pain Series**. Each episode will focus on a particular type of pain, its common causes, available treatments and future treatments on the horizon. Patient panelists and practicing experts will share their extensive knowledge and personal experiences via interactive presentations and discussions on Zoom.

Types of pain discussed will include, but are not limited to:

- **Orthopedic / Skeletal**
- **Injury-related**
- **Nerve Pain**
- **Muscle Cramping**
- **Emotional Pain**

"Pain is a multi-faceted topic and must be broken down into smaller, digestible pieces if we're going to successfully identify and treat it appropriately. No one knows your pain better than you… so, the better you are at recognizing its cause and communicating about it to others, the better your chances are of treating it".

– **Estela Lugo, CMT-Connect Moderator**

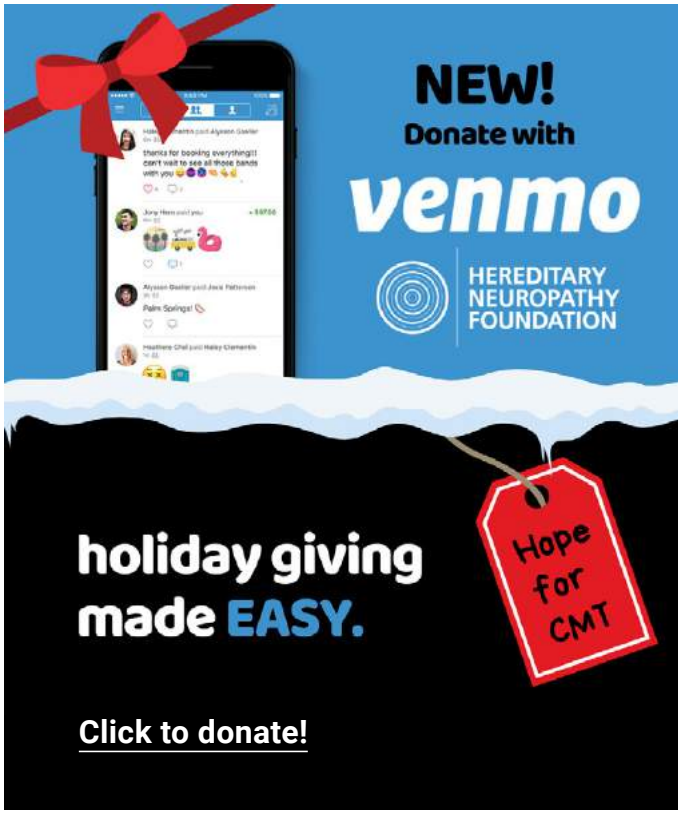
PART 1: "Does My Pain Mean I Need Surgery," featuring CMT Orthopedic Surgeon Dr. Wayne Berberian aired this month and can be viewed on the **HNF YouTube Channel**.

Stay tuned for Part 2 airing in 2021! *Date TBD

PAST WEBINARS

- Pain Series Part 1
- Guided Meditation
- Work From Home Job Training & Placement
- CMT & Telemedicine
- Align with Happiness
- CMT & Capture Proof
- CMT & Genetic Testing
- CMT & Covid-19
- Healing from the Inside Out
- CMT Resources with Inspire
- Dating & CMT
- How to Exercise in the Pool with Bernadette Scarduzio
- accessibleGO.com: A New Way to Travel with Disabilities
- Bemer Technology
- Panetta Physical Therapy
- CMT & Balance
- CMT & Your Nutrition
- CMT&Me App
- CMT & Finances
- Ability360 Sports & Fitness Center
- Active Hands
- Cannabis & CBD for CMT
- CMT & Canine Companions

VIEW PAST WEBINARS:
Click Here!



NEUROTOXIC DRUG CAUTION

To B(12) or Not to B(6)

B-Complex vitamins are essential for functioning of the nervous system and supplementation is generally well tolerated. Vitamin B12, or cobalamin, is needed to form red blood cells and DNA and is naturally found in animal foods. It can also be added to foods or supplements. B12 is a key player in the function and development of brain and nerve cells. The Recommended Dietary Allowance for men and women ages 14 years and older is 2.4 micrograms (mcg) daily.

Although rare, excess pyridoxine (B6) can result in irreversible sensory neuropathy. It is quite unlikely to reach a toxic level of vitamin B6 from food sources alone. Vitamin B6 is a water-soluble vitamin so unused amounts will exit the body through the urine. *Caution for CMT patients: Doctors may recommend supplementing with B-Complex vitamins for the treatment of chronic gastrointestinal illness or pregnancy-induced nausea, for example, but CMT patients should discuss their concerns about excess B6 with their physician prior to such treatment. The Recommended Dietary Allowance of Vitamin B6 for the general population is between 1.2mg and 1.5mg depending on age.*

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 from our site to the Directors of the HNF Centers of Excellence, Dr. Jafar Kafaie for St. Louis University School of Medicine, St. Louis, MO, and Dr. Florian Thomas for Hackensack University Medical Center & Hackensack Meridian School of Medicine, Hackensack, NJ

Q: Does neuropathy impact on the colouring of your skin e.g. after a shower, when it is cold?

A: Dr. Florian Thomas:
Skin color is impacted by many factors. Of those skin perfusion can be impacted by neuropathy via 2 mechanisms:

- The weakness in CMT of the lower leg muscles limits the movement of venous blood that has already released oxygen to the tissues to get stuck there; this can lead to ankle swelling & dark red skin discoloration. This would be relieved by lying down.
- The blood vessels themselves, in some forms of CMT can lose the nerve supply; this as well can lead to skin discoloration.

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

CMT Patient Cast in **Hallmark Christmas Movie**

This past July, HNF was thrilled to receive a call from the producer of "*The Christmas Bow*", a new Hallmark Christmas movie that premiered on Hallmark Mysteries and Movies channel on Sunday November 8, 2020 at 8pm. The part of Tess (mother of the main character) was being specially written so the character would have Charcot-Marie-Tooth (CMT) disease. Would we be able to help them find an actor with CMT in less than 30 days???

We jumped at this incredible opportunity to have an actor with CMT be front and center in a major Hallmark production. HNF was happy to announce Joy Perry was chosen longtime Team CMT supporter!

Click here to read about Joy's experience!:
https://www.hnf-cure.org/events/joy-perry-and-the-christmas-bow/

Full Showing Schedule:

Sunday, November 8	10:00 PM
Wednesday, November 11	12:00 AM
Thursday, November 12	10:00 AM
Saturday, November 14	8:00 PM
Tuesday, November 17	8:00 AM
Thursday, November 26	12:00 AM
Saturday, November 28	4:00 PM
Friday, December 4	10:00 PM
Sunday, December 6	8:00 AM
Thursday, December 10	4:00 AM
Friday, December 11	4:00 PM
Tuesday, December 15	4:00 PM
Monday, December 21	12:01 AM
Thursday, December 24	11:00 PM
Friday, January 1	4:00 AM

CMT Awareness Month — 2020, A Virtual Success
Thanks FUR Everything!

A big round of ap-paws to our furry friends and families who rallied around raising CMT awareness and research funds throughout the month of September! We are truly grateful for your 'CMT is Ruff' photos, social media frames, app downloads, educational posts, purchases, contributions and conversations. It's so encouraging to see our community come together during this challenging year to push forward with our collective mission!

We are feeling re-energized and focused for the year ahead thanks to Lisa McCarthy's Virtual 'Bold Vision' Masterclass and more connected to each other after our engaging 'CMT Movie Night' featuring 'Bernadette.' Thank you to all of our wonderful collaborators for making these meaningful events possible!

The HNF team will continue to work year-round for our friends and families living with CMT and we welcome you to connect with us for exciting new projects and events coming soon!

DONATE TO HNF RESEARCH AND PROGRAMS!
Click Here!

Young Entrepreneur with CMT1A Launches Company

Dakota Reilly is a young adult based in New York who has CMT1A. After graduating from the Fashion Institute of Technology and working in the fashion industry, Dakota realized how much she enjoys creating her own products. Since working from home is ideal for someone who has difficulty commuting, especially in New York City, Dakota decided to create her own business, Blue Bear.

Dakota launched Blue Bear in August 2020 with a full line of greeting cards, stickers and enamel pins. The products are fun and unique, with the goal of brightening your day. If you are inspired by iced coffee, plants, or a cute laptop case, you will definitely love Blue Bear. There are greeting cards for several occasions, and the stickers are weather-proof so they are great for anything you want to customize, such as a laptop, phone, or water bottle.

To check out Blue Bear's products, please visit bluebeargifts.com. Blue Bear can also be found on Facebook and Instagram at @bluebeargifts.

"Hi. I'm Alana Kohler. I'm 12 and I have CMT4A."

WRITTEN BY: **ANN L. THOMPSON**

CMT4 is a rare subtype of CMT (Charcot-Marie-Tooth disease), a genetic, neurological disorder that causes damage to the peripheral nerves — tracts of nerve cell fibers that connect the brain and spinal cord to muscles and sensory organs. It is extremely debilitating causing muscles to atrophy with severe weakness resulting in feet, legs, hands and arm deformities, sensory loss and impaired function.

"Alana was about 3 or 4 when we started to notice she walked differently," recalls her dad, Luke. "She would trip, fall, and couldn't keep up with her peers. The doctors weren't sure what was going on. Over the course of the next five, six years she was slowly deteriorating. Her actual diagnosis wasn't until 2017."

CMT4A. The "A" refers to one specific gene Alana inherited from both parents: GDAP1.

"We both had that specific recessive mutated gene," says Luke of he and Robin, Alana's parents. "There are many forms of CMT but this is basically like a one-in-a-million form. We're both healthy with very few genetic issues in either family. Robin and I went through gene testing so they could figure out if we both had the recessive mutated gene that caused the disease, and that's what it was. It was a big surprise to get that diagnosis. I couldn't believe it!"

"This gene was only discovered about ten years ago," says Allison Moore, founder and CEO of the Hereditary Neuropathy Foundation (HNF).

"After we finally got the genetic diagnosis of CMT4A, we were told that it would be many, many years before anyone looks into her type because it's so rare," says Robin. "She's had four different surgeries now. She had that big surgery where they straightened out her legs and reconstructed her flat feet, and now the latest one she had was the guided growth surgery where they actually went in and compressed the growth plates of her ankles and her knees to help straighten out her legs because of loss of muscle. The nerves don't clearly communicate with the muscles so there is muscle wasting. I mean she walks relatively well, but she has a hard time running. Definitely her braces help her. Her hands are just now starting to become affected. They told us that research would likely focus on more common types of CMT and we should just try to keep her active and keep her going because there really is no treatment. A lot of people end up completely losing function of their arms and legs and become wheelchair dependent."

"It's a hard disease to have," says Alana. "Sometimes I feel embarrassed in front of my classmates. It's not easy to keep up. The hardest part is the bullying. It makes me feel really sad that I'm not, like, normal and I feel like I shouldn't have CMT4A. Why me? I just try and stay strong."

"A lot of society is based on looks, the way you walk, the way you talk, who your friends are, what sports you play," says a resigned Luke. "A lot of her friends growing up were playing different sports but they didn't take her along. Seeing her struggle with that, not being able to, I guess, fit in to some of those crowds or people, and then struggling with constant teasing and being made fun of at school…"

Her big, warm smile and generous heart can melt metal. She used to dance.

Luke takes a deep, slow breath. "That's the saddest part."

Any sufficiently advanced technology is indistinguishable from magic.

— Arthur C. Clarke, science writer & author

"We joined a bunch of Facebook support groups including the Hereditary Neuropathy Foundation," recalls Robin. "A couple of months ago they put out a call for all CMT4A families to join this group and we learned of a research project. About twenty families from around the world are going to participate. They hope to have a treatment within the next two to three years that would halt the progression and possibly even help Alana regain some of her lost function. Alana is super excited about all of this! It's the miracle that we've been praying for. We're very hopeful."

"Scientists have developed a new gene-editing technology that could potentially correct up to 89% of genetic defects, including those that cause diseases like sickle cell anemia."

— Jessie Yeung, CNN, 2019. Gene replacement, a variant of gene-editing, looks like a promising treatment for CMT4A.

Alana and her family have become a part of a revolution in modern medicine. Some would call it a miracle. Dr. Mario Saporta, MD, a specialist in neurology and neuromuscular medicine at the University of Miami explains that "gene therapy is very specific to the gene you are targeting. In this case, it's in the DNA region called GDAP1. There are two different diseases caused by the same gene, CMT2K and CMT4A." Alana's version, 4A is "very amenable for gene replacement therapy because the gene she inherited isn't operating. It's the loss of the gene that is causing the disease. Replacement will resolve the problem."

October 8, 2012: The Nobel Assembly at Karolinska Institutet: "The Nobel Prize recognizes two scientists who discovered that mature, specialized cells can be reprogrammed to become immature cells capable of developing into all tissues of the body. Their findings have revolutionised (sic) our understanding of how cells and organisms develop."

— www.nobelprize.org

Referencing a 2012 Nobel Prize winning development, Dr. Saporta explains, "We can take blood or a small piece of skin from the patient and by manipulating those cells we can turn back their biological clock and turn them into a stem cell state. (A stem cell is a cell that isn't programmed—a clean slate so to speak.) We call those iPSC (induced Pluripotent Stem cells). It's Revolutionary! By looking at what's wrong with that cell, we can look to see if we can correct it by using a gene replacement therapy, or a compound, or a drug. It's almost like plug and play. It's basically just changing out the gene." He makes it sound so easy!

"It was the best day ever when we learned of that gene therapy research project a couple months ago," a joyful Robin says. "It's given us so much hope that there is actually potential for a cure! Alana could potentially live a life with fewer physical challenges. It's so hopeful."

There's still lots of work to be done before gene replacement is available to Alana. "We do have to go through the process of showing the efficacy and safety, but things become a little easier each time we are successful applying those principles," says Dr. Saporta. "We've seen this being successful with other genetic approaches. This has enabled us to be way more targeted and faster. I think this is the snowball effect. Every successful case moves us faster and forward."

Of course there's a "but…" Isn't there always? "The challenge," explains Allison (of HNF), "is money. It'll take $3 million to get us to human trials. There is urgency here. Alana is young and, depending on when we can raise the money, this disease might be halted in Alana and there's even a possibility that some functionality could be restored."

This is where you can help make magic or, if you prefer, miracles:
Donate to GDAP1: Click Here

"The more money we raise, the faster we can get to the clinic," Allison says excitedly. "Money earmarked for CMT4 can only be used for this mutation. Traditionally, we fundraise through direct gifts and families sponsoring events. COVID slowed us down like everybody else."

"It's just kind of amazing that we're at a point where we could halt the progression," says an encouraged Luke. "If we could just keep her the way she is right now and maybe, hopefully, potentially getting her little better, it's like man, it doesn't get any better than that!"

"Honestly, if I didn't have this, I'd be a huge runner or long walker," says Alana. She might even dance again.

Never doubt that a small group of thoughtful committed citizens can change the world. Indeed, it is the only thing that ever has.

— Margaret Meade

Donate to GDAP1: Click Here

New Pediatric Center of Excellence: Children's Hospital Colorado

DR. MICHELE YANG

Q: TELL US ABOUT YOURSELF:

I am a board certified child neurologist with subspecialty training in electromyography and neuromuscular disorders.

Q: WHY IS CMT YOUR PASSION?

CMT is a complex group of disorders both clinically and genetically. In the last decade our understanding of the genetics and pathophysiologic mechanisms has grown tremendously, and it has been exciting to see how this understanding may lead to treatments for patients with CMT. Because the clinical presentations of CMT can vary, I look at how CMT uniquely affects the child and family. In our multidisciplinary clinic with rehabilitation medicine specialists, orthopedic surgeons, physical therapists, occupational therapists and genetic counselors, we work together to improve the care for our patients. I believe this team approach to care is best for CMT patients, and I truly enjoy working with this wonderful group of providers.

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

When patients come to our clinic, they will first meet our nurse, Lori Yacone, and our coordinator, Alison Ballard. They are the point of contact for our patients should any issues arise. If they are new patients, they will meet the neurologist and the genetic counselor, who will review any diagnostic workup needed to make a diagnosis of CMT, including genetic testing. The genetic counselor will provide counseling to families who are new to the diagnosis of CMT and will assist with testing of other affected family members if requested. At every visit, patients will meet with the rehabilitation specialist, orthopedic surgeon, and occupational and physical therapists. A neuropsychologist is also part of the team, and will see patients who have any concerns about learning issues that may arise.

Q: WHAT DO YOU LOVE MOST ABOUT YOUR PRACTICE?

I love seeing the children in the clinic grow into wonderful adults. The providers I work with in the clinic are wonderfully caring people passionate about the care of patients with neuromuscular disease. I feel fortunate to work with them.

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

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

HNF's New Center of Excellence: AtriumHealth Charlotte, NC

DR. URVI DESAI

Q: TELL US ABOUT YOURSELF:

I am a board certified neurologist with added board certification in neuromuscular disorders and EMG. I see patients across different age groups with varied neuromuscular disorders, be it genetic, autoimmune, inflammatory or degenerative. I direct a large comprehensive multidisciplinary neuromuscular clinic at Atrium health supported by an AANEM accredited EMG lab. I love to read, listen to Indian classical music and travel the world with my husband. I have a daughter who lives in New York City.

Q: WHY IS CMT YOUR PASSION?

CMT can affect patients since their younger years and can have a significant impact on quality of life caused by weakness, numbness, imbalance and neuropathic pain. Many patients suffer disabilities that can affect them profoundly. The genetics of CMT is also quite complex and yet there is no treatment available for this disease. However, there is more research going on to find a cure and to understand the pathophysiology of this disorder. I have been involved in clinical trials in CMT patients and am looking forward to offering more options to our CMT patient population. I am honored that our clinic is recognized as an HNF Center of Excellence.

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

We provide a large multidisciplinary clinic comprised of neurologists, physiatrists, nutritionists, social workers and physical, occupational, respiratory and speech therapists. The clinic also helps to coordinate services of equipment specialists for repair and delivery of assistive devices. In addition to that, we work closely with other specialties like cardiology, pulmonary and genetics. The clinic aims at providing appropriate care for our CMT patients in a wide-ranging multidisciplinary fashion and also offers options to participate in clinical trials and research.

Q: WHAT DO YOU LOVE MOST ABOUT YOUR PRACTICE?

I love the ability to provide advanced care to my patients in our multidisciplinary clinic. In addition to that, patients also have access to participation in clinical trials and research activities. There has been significant progress in diagnosis and treatment of neuromuscular disorders like CMT in the last few years and I love to be at the forefront of this advancement to be able to help my patients.

Atrium Health Neurosciences Institute-Charlotte
1010 Edgehill Road North
Charlotte, NC 28207

The phone number for appointments is 704-446-1900. Ashley Clyburn is our incoming referrals coordinator and referrals can be faxed to 704-355-5650.

HNF's New Center of Excellence: Department of Neurology at UCLA

DR. ANASHEH HALABI

Q: TELL US ABOUT YOURSELF:

I'm an Assistant Clinical Professor in the Department of Neurology at UCLA where I specialize in neuromuscular diseases. Growing up, I was certain I'd be an astronomer. I was fascinated by the vastness of space and our place in the universe. My favorite childhood memories are of being at the Griffith Observatory at night. As I got older, I realized so much of what fascinated me about space is reflected in the brain, a network of atoms from which we create music and art.

After completing my undergraduate degree in neuroscience and music history at UCLA, I moved to New Orleans to pursue my MD and PhD in Genetics at Louisiana State University. In my clinical years, I had the opportunity to spend time in the New Orleans Musicians Clinic with my mentor, Dr. John England. It was here that I realized how critical it was to translate scientific discovery into patient care. It matters not just to understand how someone's neuropathy may affect their dexterity from a scientific standpoint—it matters how we express these discoveries to patients, and how we manage their care. Loss of dexterity could mean loss of livelihood or loss of feeling alive when you put your hands on a piano.

In 2015, I returned to Los Angeles where I completed my internship at Cedars-Sinai Medical Center and then moved to San Diego for my Neurology Residency where I was also Chief Resident. Those years honed my love for teaching and academic medicine. In 2019, I came home to UCLA for my neuromuscular fellowship and I am thrilled to begin this next chapter in my career developing a new vision for clinical care in academic Neurology.

Q: WHY IS CMT YOUR PASSION?

I would argue that CMT patients are my passion – they are categorically unstoppable. It is incredibly inspiring to watch CMT patients lead the charge and advocate for themselves. To that end, with the rapid development of new therapies in other areas of nerve and muscle diseases, my hope is to be a part of that charge and movement towards better access to structured, streamlined care.

Q: WE UNDERSTAND YOU ARE BUILDING A STATE OF THE ART MULTIDISCIPLINARY CENTER AT UCLA. WHAT MAKES UP A MULTIDISCIPLINARY CLINIC?

So often after the diagnosis is made, CMT patients are piecing together their care and information about expectations on their own. The dream is the development of a comprehensive

Department of Neurology at UCLA

program that would allow patients to have a multi-disciplinary experience when they come in for a visit. In this initial phase, as I aim to identify means by which to fund this kind of endeavor, the goal is to target and invest in the patient's greatest needs first and grow from there. With UCLA's breadth of resources, including allied health professionals and clinicians in other areas, I can act as a conduit and eventually, we can operate in parallel, as a one-stop-shop.

Q: WHAT WILL PATIENTS EXPERIENCE AT THEIR FIRST VISIT, HOW LONG CAN THEY EXPECT TO BE THERE AND WHAT TECHNOLOGIES OR SERVICES WILL YOU OFFER THEM?

Right now when patients come into the clinic, I do a comprehensive consultation with a neurologic exam to establish whether or not we have a meaningful explanation for their symptoms (a diagnosis) and offer access to an orthotist, referrals to physical therapy, and other programs in the region. This initial visit will last about an hour.

The COVID-19 pandemic has led to an acceleration of access to telehealth within our system. Should there be physical limitations in getting to Los Angeles, we have a multitude of ways in which we can connect with patients.

Q: DO YOU SEE ADULTS AND PEDIATRIC PATIENTS?

Yes absolutely, all are welcome!

Q: WHAT DO YOU LOVE MOST ABOUT YOUR NEW ROLE AS DIRECTOR AT UCLA?

Beginnings are exciting opportunities for growth and hope. My dialogue with the Hereditary Neuropathy Foundation has truly been one of my favorite parts of this endeavor!

Q: HOW CAN PATIENTS MAKE AN APPOINTMENT AT UCLA DEPARTMENT OF NEUROLOGY AT UCLA?

UCLA Department of Neurology
300 Medical Plaza, Suite B200
Los Angeles, CA 90095

Dr. Halabi Anasheh Office
Telephone: 310-794-1195
(note: request to be scheduled with Dr. Halabi)
Fax: 310-794-7491

HNF's CMT Centers of Excellence

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

ARKANSAS

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

CALIFORNIA

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

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

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

COLORADO

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

CONNECTICUT

Hospital for Special Care
New Britain, CT
Contact: 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: Mrs. Molly Labrier
314-977-6177
molly.labrier@health.slu.edu

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

NEW JERSEY

Hackensack University Medical Center
Hackensack, NJ
Contact: Florian Thomas, MD, PhD
551-996-8100
Annerys.Santos@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
Contact: For clinical appointments
Allan Paras 212-305-0405
For research studies 212-305-6035
ap3476@cumc.columbia.edu

NORTH CAROLINA

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

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

TEXAS

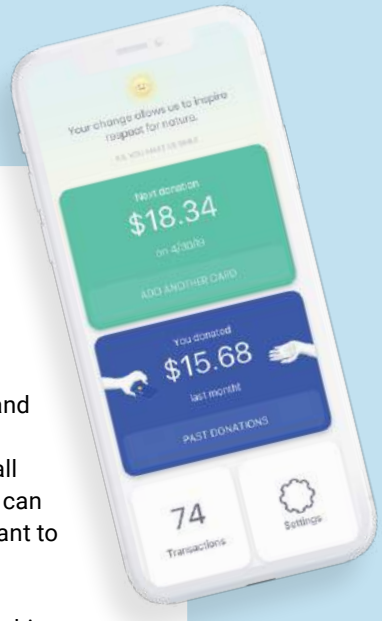
Austin Neuromuscular Center
3901 Medical Parkway, Ste. 300
Austin, TX 78756
Contact: Yessar Hussain, MD
512-920-0140
Website: austinneuromuscle.com/contact

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

HNF
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New York, NY 10016
hnf-cure.org

Events

Get Involved and Join us at an Upcoming HNF Event

Virtual Movement is Medicine™ Summit

March 7, 2021
Winter Park, Florida

Join us for a virtual day of CMT-friendly exercise classes and workshops. Registration opening in January 2021.

Movement is Medicine™ Summit

November 5-6, 2021
Phoenix, Arizona

Join us for our 2nd Movement is Medicine™ Summit
Details will be released soon!

Jaxson's Crusaders Spooky Clay Shoot

On Saturday October 31, 2020, the Flynt Family and their close friend Heather Guerrero held the Jaxson's Crusaders Spooky Clay Shoot to raise money for gene therapy research. The Flynt's son, Jaxson, was diagnosed with Leigh syndrome C12orf65 five years ago and they have been raising money for research ever since. The family-friendly event included a bounce house, face-painting, a trunk-or-treat and kids' games. The clay shoot had over 144 shooters and around 175 total participants raising $35,000. They are looking forward to their next charity clay shoot in Fort Worth in October 2021.

Donate to Jaxson's Crusaders: Click Here!

PHARNEXT

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