HEREDITARY NEUROPATHY FOUNDATION

JANUARY-MARCH 2017

cmtupdate

New Collaboration with the University of North Carolina at Chapel Hill and The Jackson Laboratory Addresses Charcot-Marie-Tooth/Inherited Neuropathy Mutations Using Gene Therapy

BY **SEAN EKINS,** CHIEF SCIENCE OFFICER, HNF

Gene therapy represents an exciting new frontier being explored for treating a number of diseases. Currently, gene therapy is being used in a clinical trial to treat one of the rare forms of inherited neuropathy (IN), giant axonal neuropathy (GAN). Because of the potential gene therapy holds for treating a range of CMT/IN mutations--including the rarer ones—HNF is excited to announce a collaboration with the University of North Carolina at Chapel Hill and The Jackson Laboratory to begin the development of gene therapy approaches for Charcot-Marie-Tooth (CMT) and Inherited Neuropathies (IN).

To start, our initial gene therapy work will focus on CMT type 6, which is caused by a recessive mutation in the C12orf65 gene and for which there is currently no treatment. CMT type 6 presents in patients with many of the typical symptoms associated with most types of CMT, but usually includes additional, significant impacts on vision and breathing that further challenge a patient's quality of life. And, unlike other forms of CMT, CMT type 6 is ultimately fatal.

The CMT type 6 research that HNF is funding aims to replace the non-functional gene with a working copy. Dr. Robert Burgess, lead co-investigator for this project, has developed a CMT type 6 mouse model to conduct a proof of concept study to determine the viability of using this approach to treat the disease. Dr. Steven Gray, assistant professor at the University of North Carolina, will be developing the gene therapy vector to be tested using this unique mouse model. We are pleased to have Dr. Gray on this project's team as his previous work was responsible for bringing the GAN gene therapy to a clinical trial.

Ensuring the success of this gene therapy approach will depend on getting everyone—health care providers and CMT type 6 patients and their families—involved! Besides the work in the CMT type 6 mouse, patient specimens are

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1-855-HELPCMT (435-7268)

www.hnf-cure.org

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.

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GENE THERAPY RESEARCH

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extremely valuable for research. HNF will need to obtain skin fibroblasts from CMT type 6 patients that have a confirmed diagnosis. HNF will rely on its Centers of Excellence network and other medical institutions to obtain the skin fibroblasts needed to be sent to The Jackson Laboratory for further testing.

Advancing a research project like this is a major undertaking requiring significant human, physical, and financial resources—however, the possibilities of achieving significant outcomes are unprecedented. To secure the ability of this project's momentum, HNF has launched the Gene Therapy Fund to support this project's efforts to tackle additional types of CMT/IN that warrant a gene therapy approach to treat CMT. The Gene Therapy Fund will be used and supported with the collaboration of Lori Sames (co-founder of Hannah's Hope Fund and the tireless champion of the first gene therapy ever used for any form of IN), UNC and The Jackson Laboratory.

We need your help! Here's how to get involved:

I support gene therapy research, and I want to enroll as a patient!

If you are a patient with CMT and want to be part of this new initiative, join the following programs to ensure you are considered for participation:

1. **Global Registry for Inherited Neuropathies (GRIN)** If you have any type of CMT/IN or do not know your type, join here: CLICK HERE
2. **Charcot-Marie-Tooth Research Network (CMTRN)** If you have HNPP, CMT2A with visual impairment/optic atrophy, CMT2C, LMNA mediated AD-CMT2, CMT4, CMT4A, 4B1, 1B2 & 4B3, 4C, 4D, 4E, 4F, 4G, 4H, 4J, CMT6, Autosomal Dominant Optic Atrophy ADOA (OPA1) and Giant Axonal Neuropathy (GAN). To join: CLICK HERE

I want to have my mutation considered!

If you are interested in having your mutation added to the list or in championing a gene therapy approach for your type of CMT/IN, please contact Allison Moore at allison@hnf-cure.org

I support gene therapy research, and I want to make a donation!

Your donations are crucial to making this research possible.

PLEASE DONATE TODAY TO HNF'S "WHAT IF?" GENE THERAPY FUND TO SUPPORT THIS GROUNDBREAKING RESEARCH AT: CLICK HERE

Tina Tockarshewsky Joins HNF Team to Increase Outreach!

We are pleased to announce that Tina Tockarshewsky has joined the HNF team in the newly created role of Director, Medical and Public Affairs. Tina will help HNF with its outreach to the health care provider community—including our Centers of Excellence program—as well as increase our advocacy efforts to promote greater awareness of the challenges our CMT community faces.

Some of you may recognize Tina's name already as she previously served as president and CEO of The Neuropathy Association. A highly-respected patient advocate for peripheral neuropathy and nerve pain, her efforts led to her appointment on significant Federal chronic pain committees. She has been an active presenter at professional and general public health care forums, and her outreach on behalf of the Association's community elevated the national profile for all forms of neuropathy and promoted a greater awareness of the challenges in treating neuropathic pain. Tina's recent public service has included:

- Member, Interagency Pain Research Coordinating Committee (HHS Appointment);
- Contributor, National Pain Strategy Task Force-Public Education & Communications Working Group;
- Executive Committee Member, Analgesic Clinical Trials Translations, Innovations, Opportunities, & Networks (ACTTION) executive committee member (FDA private-public partnership initiative); and,
- Contributor, Initiative on Methods, Measurement, & Pain Assessment in Clinical Trials (IMMPACT)

Most recently, Tina has been involved in consulting work focused on developing patient engagement strategies and patient community outreach campaigns. In 2015, she authored a cover article for *BioSupply Trends Quarterly* magazine on "The Perfect Storm for Patient-Focused Clinical Trials," which looked at the needs of patient communities vis à vis the changing winds of policy development and R&D efforts seeking to be more "patient-centric."

We are delighted to have Tina on board, especially at this time of increasing momentum for our community's efforts. Please join us in welcoming Tina to her new role!

TRIAD
THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY

HNF developed the Therapeutic Research in Accelerated Discovery (TRIAD) as a collaborative effort with academia, government and industry, to develop treatments for CMT. Currently, TRIAD involves many groups that span the drug discovery, drug development, and diagnostics continuum.

BY **SEAN EKINS**, CHIEF SCIENTIFIC OFFICER, HNF

HOT OFF THE PRESS!

1.

The most common form of axonal CMT is called CMT2A and it is caused by mutations in mitofusin-2 (MFN2). Nerve biopsies from CMT2A patients show axon loss and occasionally abnormal Schwann cell morphology and mitochondrial groupings (the cell's powerhouse). In order to understand the disease and potentially develop treatments we need an animal model that replicates CMT2A in humans. A pilot grant from HNF was funded Dr. David Pleasure (Shriners Hospitals for Children and University of California, Davis) to develop a mouse model. They chose to knock in the most prevalent MFN2 mutation in the U.S., namely T105M, to give hemizygous expression of MFN2. They then characterized the muscle/nerve interactions. The mice were shown to have abnormal myelination and aggregation of mitochondria in the Schwann cell cytoplasm. A statistically significant decreased hind limb footprint length versus wild type was reminiscent of pes cavus foot deformity in CMT2A patients. This study provides a starting point for a new animal model already shared with The Jackson Laboratory, where Dr. Robert Burgess will see if a more pronounced phenotypes can be developed that can, in turn, be shared with the research community. We hope that these will then be used to help academic and industrial scientists as they search for treatments to reverse CMT2A.

2.

Mitofusins (MFN) are important for tethering and fusion of the mitochondria outer membrane, which is critical for development in an embryo. Mutations in MFN2 interrupt this ability to fuse and can cause neurodegenerative diseases like CMT2A. Dr. Gerald W. Dorn II (Washington University School of Medicine, St. Louis) and colleagues have modeled the structure of MFN2 and determined that it exists in different conformations. One can imagine the structure as a coil in a closed state with different regions interacting to hold it in place, which then opens it to extend for mitochondrial tethering. When there are mutations, this opening is prevented. They also used minipeptides—which can be considered as small pieces that are complimentary to MFN2 and which compete with the internal interactions, thus altering the conformation of MFN2. This was proposed as a potential way to move to their active extended conformations to overcome the mutations in MFN2 that result in CMT2A. This approach was shown to work in cultured mouse neurons, reversing the mitochondrial pathology, which suggests that controlling MFN2 unfolding to promote mitochondrial tethering is an alternative to gene therapies. What remains to be seen is how a viable minipeptide could be delivered in animals or humans and whether it would be able to reach its target. In the meantime, though, it represents a useful proof of concept.

TO SUPPORT RESEARCH FOR CMT2, MAKE A DONATION TO THE THE HELP FUND: CLICK HERE

Calling Everyone With CMT/IN

CMT/Inherited Neuropathies Patient-Reported Survey Needs You to Advance Therapies

HNF has launched a new Patient-Reported Research Study to advance therapies for CMT and inherited neuropathies (IN) in an effort to collect comprehensive data for a better understanding of the disease. The study aims will result in the most comprehensive understanding of the disease and help better characterize and describe the CMT/IN patient population. The study's eligibility is open to all types of CMT/IN and, as a whole, will enhance the understanding of the severity of symptoms, characteristic signs (pes cavus, poor or absent reflexes), genotypes, treatments, and patients' willingness to participate in research and in clinical trial design. This information will assist in the development of recommendations and standards of care guidelines, as well as be useful for identifying clinical endpoints, patient-reported functional outcomes, and best design approaches for clinical trials.

This web-based survey is in an online questionnaire format with multiple choice answers that can be checked off by the participants, with some questions offering the option to fill in an answer when applicable. There is no identifiable personal data, just detailed information that will be collectively analyzed for research purposes and shared within the CMT/IN stakeholder community. HNF will use this survey to continue to build upon the findings that were gathered during the recent 2016 Patient-Centered CMT Summit, which was partially funded by the Patient-Centered Outcomes Research Institute (PCORI).

TAKE SURVEY AT: CLICK HERE

TRIAD

Natural History Study to Support CMT Research

The National Organization for Rare Diseases (NORD)—an independent charity that built its natural history study platform for rare diseases and is supported by a cooperative agreement by the U.S. Food and Drug Administration (FDA)—has awarded HNF funding to build a robust natural history study to support the second most common type of CMT called hereditary neuropathy pressure palsies (HNPP).

This study will also include the rarer types of CMT including HNPP, CMT2A with visual impairment/optic atrophy, CMT2C, LMNA mediated AD-CMT2, CMT4, CMT4A, 4B1, 1B2 & 4B3, 4C, 4D, 4E, 4F, 4G, 4H, 4J, CMT6, Autosomal Dominant Optic Atrophy ADOA (OPA1) and Giant Axonal Neuropathy (GAN). These rarer types often have unique and severe symptoms that may be fatal.

HNF is proud to be one of twenty leading rare disease groups selected for this NORD initiative. HNF sees great opportunity in partnering with NORD to fill the research gaps that can lead to a better understanding of how these rare diseases progress over time.

Natural history studies are imperative in our understanding of rare diseases like CMT. Identifying the more unique symptoms among CMT patients—such as optic atrophy (including vision impairment and blindness), hand tremors, vocal cord paresis, hearing loss, and cognitive deficits—can produce the vital information needed for clinical trial design.

Currently, there is no cure for CMT/IN, but clinical trials for the most common type CMT1A are underway.

Allison Moore, HNF founder and CEO—who herself has CMT1A, as do several members of her family—emphasizes the need to turn our attention to the rarer forms of CMT:

"For the rarer forms of CMT, which often have very severe symptoms, there is less hope for treatments and cures. I am passionate about ensuring that all types of CMT/IN have pipeline drugs and gene therapies to support the patient community. Collecting patient-reported information will be invaluable and will be made available to any researcher or drug or gene therapy developer interested in creating therapies for CMT."

The new Natural History Study is part of HNF's Charcot-Marie-Tooth Research Network (CMTRN), enabling us to provide a complete picture of each patient's experience with CMT/IN. HNF is launching this initiative to help identify patients with HNPP and the more rare forms of CMT/IN.

To date there is very little data on the rare forms of CMT/IN, and finding patients all over the world is critical in therapy development. If you are a patient or a family member (or know someone with CMT/IN), please join this critical study.

The goal is to enroll as many patients as possible. Joy Aldrich, HNF advocacy director and moderator for HNF's Inspire Online Support Group, states:

"The success of this natural history study is dependent upon community participation: we need all CMT/IN patients to participate."

Debi Houliares, community advocate, HNF board member, and mother of a son with a rare form of CMT adds:

"As the mother of a son with CMT6, it is so exciting to see our community having access to a study that not only includes the rarer forms of CMT, but enables us to track these diseases over time to identify and validate the daily physical and quality of life challenges we know we are struggling with — but have yet to have had a way to document and quantify these challenges in any sort of productive, scientific way for future research."

The CMTRN supports natural history studies via electronic surveys that collect patient experience and disease progression information. Patients (or their caregivers or guardians) can enter information from anywhere in the world. The data is made anonymous and stored securely in an online portal. HNF can share the data with individuals or institutions conducting research or clinical trials, but cannot share any personally identifying information. This format is approved by the study's governing board, which includes scientists, healthcare providers, and patient advocates.

NORD president and CEO Peter L. Saltonstall says:

"NORD's natural history studies platform empowers patients and families to drive research and eliminate some of the unknowns that still exist in rare diseases. We are glad to be working with the Hereditary Neuropathy Foundation, one of our member organizations, on this project, and we thank the FDA for its support and on-going commitment to help people with rare diseases."

HNF is excited to be a part of NORD's Natural History Studies Project.

BE SURE TO SIGN UP SO WE CAN MAKE THIS STUDY AS EFFECTIVE AS POSSIBLE AND HELP ENCOURAGE OTHERS TO PARTICIPATE AS WELL!
CLICK HERE

PARTICIPATE TODAY:

ADULTS WITH RARE DISORDERS SUPPORT STUDY

HNF had the opportunity to connect with Kathleen Bogart, PhD, the principal investigator of the "Adults with Rare Disorders Support Study" in partnership with the National Organization for Rare Disorders (NORD). She is an assistant professor of Psychology at Oregon State University, where she studies the psychosocial needs of people with rare disorders and has a rare disorder herself. Dr. Bogart also serves on the board of directors of a NORD member organization.

What is the study about?

The "Adults with Rare Disorders Support Study" is the first large-scale study about the information and psychosocial support needs of people living with rare disorders. The study will assess these needs, from the perspectives of patients with a variety of rare disorders, to find similarities and differences across disorders.

It's crucial for as many people living with a rare disorder participate in this study to accurately reflect the diversity within the rare disorder community. Data can be published with 60 study participants per disorder.

How do patients participate in the study?

There are two ways to participate:

1. Patients can take a 40-minute online survey about their experiences, providing information and support needs related to their rare disorder (paper forms are available by request). If it is physically difficult to respond, someone may enter responses for the participant.
2. During the survey, participants can opt to sign up for a second study. This study involves an online focus group (video, not in person) about the information and psychosocial support needs with others with rare disorders. Participation in the survey is required in order to be eligible for the focus group, but the focus group study is not required to participate in the survey. Focus group members will be paid $20 for participation.

Who is eligible to participate?

In order to participate, patients must be 18 or older, be able to communicate in English, and have a rare disease or disorder or undiagnosed rare condition. **Caregivers who do not have a rare disorder themselves are NOT eligible to participate at this time.**

Disorders are generally considered rare if it affects fewer than 200,000 affected individuals in the United States, or fewer than 1 in 2,000 in Europe.

Because rare disorders are discovered and prevalence estimates change frequently, patients may participate even if their disorder does not appear on the list.

What will be done with the study findings?

A summary of results will be sent to all participants. To help NORD, rare disorder organizations, and healthcare professionals meet the needs of people with rare disorders, results will be shared through reports, conference presentations, and scientific publications.

Who are the researchers?

Dr. Bogart, PhD will be joined by co-investigator Veronica Irvin, PhD, MPH, an assistant professor of Public Health at OSU. She has experience analyzing information offered by support organizations.

PARTICIPATE AT: CLICK HERE

FOR MORE INFORMATION ON THE STUDY, CONTACT:
KATHLEEN.BOGART@OREGONSTATE.EDU OR 541-737-1357.

A LIST OF RARE DISORDERS CAN BE FOUND AT:
CLICK HERE

Inaugural Patient-Centered CMT Summit

Demonstrates the Power of Patient-Reported Outcomes

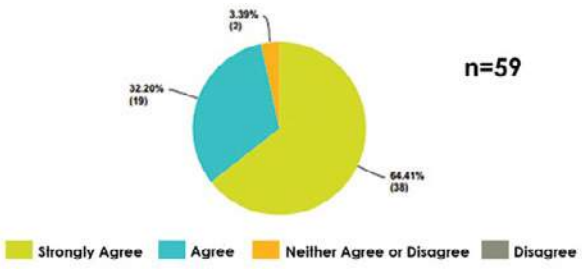
BY **ALLISON MOORE,** FOUNDER & CEO/PRINCIPAL INVESTIGATOR, HNF

On October 6, 2016, HNF held the first-ever Patient-Centered CMT Summit in New York City. Funded in part by a Eugene Washington PCORI Engagement Award and designed by a patient-driven planning committee, this well-received one day conference offered expert sessions and primarily patient-led panels focused on patient engagement methods: all with an emphasis on identifying the patient-reported outcomes (PROs) gaps that are hindering patient care, standard of care guidelines, diagnosis, and therapy development. With 166 participants representing all facets of the CMT/inherited neuropathy (IN) community, Summit discussions focused on care and research priorities most meaningful to patients, all in the hopes of encouraging the utilization of PROs in patient-centered outcomes research/comparative effectiveness research (PCOR/CER). Patient attendees reported that the Summit helped them feel less isolated, with some meeting another person with CMT/IN for the first time. Patients and their caregivers appreciated having one-on-one interactions with expert clinicians for genetic testing guidance, to learn about possible symptom management treatments, plus to hear panel discussions on topics specific to CMT families. Researchers observed that interacting with patients gave them new insights on ways to better translate their research. Industry participants were also engaged by seeing first-hand patients' commitment to advocacy and enthusiasm for research and clinical trials participation. Our on-site survey generated insights into new ways industry could improve clinical trial design when thinking about PROs related to falls, as well as demonstrated patients' tolerance for trial site visits (including visit duration, frequency, distance).

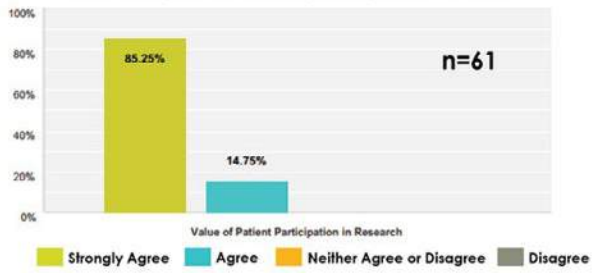
The event also revealed that much work still needs to be done to address patient needs. Gaps in patient and health care provider (HCP) knowledge and awareness were identified as hindering diagnosis and the delivery of proper treatment options. Patients consistently reported years of frustration prior to being diagnosed, followed by disappointment in the lack of guidance from their HCPs regarding on-going disease management. Attendee surveys showed strong evidence that patients and health care providers have not been collaborating. Collaboration can tremendously impact standard of care guidelines, clearer understandings of patient participation in research, and therapy development through the use of patient-reported outcomes. The Summit's survey shows that patients could benefit immediately from better management of their disease using currently available resources. With help from knowledgeable clinicians, patients could already obtain earlier and more accurate diagnosis, receive proper care for symptom management by being informed about options such as surgery, leg braces, physical and occupational therapy, and can also be made aware of clinical trials. HNF thanks PCORI and all of the participants who made this unique Summit possible. The event has already initiated new research protocols to bridge the gaps identified related to PROs and treatment options. We look forward to continuing to close the gaps to improve the lives of CMT/IN patients.

CARE AND RESEARCH GAPS IDENTIFIED

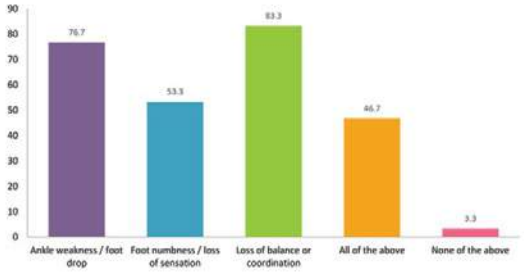
Do you feel more connected to the patient community?



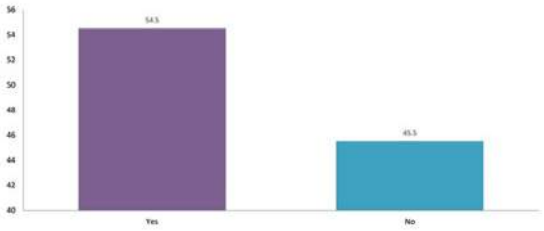
Do you see the value in patients participating in the research process to improve patient care?



What are the factors that contribute to your risk of falls?



Have you been genetically tested for CMT or inherited neuropathy?



PATIENT-CENTERED CMT SUMMIT: CLICK HERE **PATIENT-CENTERED OUTCOMES RESEARCH INSTITUTE (PCORI):** CLICK HERE

Charcot-Marie-Tooth (CMT) Disease Treatment Advances Propelled Forward by Diagnosis and Patient Engagement

CALL TO ACTION!

BY **TINA TOCKARSHEWSKY**, DIRECTOR OF MEDICAL AND PUBLIC AFFAIRS, HNF

HNF is a staunch champion for accurate and early CMT disease diagnosis, recognizing its critical importance for patient care and research. CMT encompasses a heterogeneous group of inherited, progressive, chronic peripheral neuropathies. CMT type 1A (CMT1A), the most common type, is an orphan disease affecting at least 125,000 people in Europe and the U.S. For those with CMT1A and suffering with its progressive muscle atrophy in their legs, feet, arms and hands, there are currently no cures nor approved curative or symptomatic medications.

A change may be on the horizon, though: in December 2016, Pharnext, a French biopharmaceutical company, completed patient enrollment for its international Phase 3 PLEO-CMT trial of PXT3003, Pharnext's lead PLEODRUG©, for CMT1A treatment. PLEO-CMT is a multi-center, randomized, double blind, placebo-controlled, adaptive design Phase 3 study initiated in December 2015, with 323 patients with mild to moderate CMT1A enrolled in 30 sites across Europe, the U.S. and Canada. The trial's primary endpoint is the change in the Overall Neuropathy Limitation Scale (ONLS) to determine improvement of patients' disability after 12 and 15 months of treatment. Trial results are expected in the second half of 2018. PXT3003 is a novel oral fixed-low dose combination of (RS)-baclofen, naltrexone hydrochloride and D-sorbitol. The exploratory Phase 2 trial demonstrated safety, tolerability and improvements beyond stabilization of CMT1A patient disability (Orphanet Journal of Rare Diseases http://www.ojrd.com/content/9/1/199). HNF has been partnering with Pharnext to encourage this clinical trial's development, and we want to thank all who are involved with the study, from the researchers and developers at Pharnext, the clinicians in the field and at our HNF-designated Centers of Excellence, to the patient participants—all contribute and play a critical role!

Two factors are critical in moving a trial such as this forward: patients getting an accurate CMT1A diagnosis and then finding/enrolling these patients. If this therapy becomes available, patient access will also depend on an accurate diagnosis; for many, this is still a significant gap. A patient poll conducted at HNF's 2016 Patient-Centered CMT Summit showed only 54.5% of respondents had been genetically tested for CMT or an inherited neuropathy (IN); 61.1% said they knew what type of CMT/IN they actually had.

Patient data drives research: HNF encourages health care providers to have their patients join HNF's Global Registry for Inherited Neuropathies (GRIN) -www.neuropathyreg.org. GRIN seeks to identify patients to gain a better understanding of disease phenotypes and patients' willingness for clinical trial participation. GRIN's data includes patient-reported outcomes and aims to generate data that could close the knowledge gaps regarding disease progression and disease experience.

The CMT/IN community has unprecedented opportunities within its grasp: active participation by all stakeholders and greater awareness of CMT/IN resources (diagnostic tools, clinical trials, patient registries and networks) can encourage therapeutic and standards of care improvements. Stay engaged with HNF, encourage patient involvement...and stay tuned!

"We have been pleased to contribute to this groundbreaking research by providing U.S. clinical site identification and patient recruitment support for this pivotal Phase 3 trial of PXT3003," shares Allison Moore, HNF founder and CEO. "HNF resources such as the Global Registry for Inherited Neuropathies (GRIN), our online Inspire Patient Community and our CMT-Connect local patient empowerment programs have been instrumental. We are enthusiastic that our joint effort with Pharnext could result in providing much-needed new therapies, and we look forward to our joint work for years to come."

PHARNEXT

CALLING ALL PEDIATRIC NEUROLOGISTS!

New clinical trials are underway to treat Charcot-Marie-Tooth Disease.

Your input is needed to gain a better understanding of clinical endpoints and outcome measures for children living with CMT.

TAKE OUR QUICK SURVEY AT WWW.HELPCMTKIDS.ORG

The Hereditary Neuropathy Foundation (HNF) is committed to improving clinical endpoints and functional outcomes in the pediatric patient population with Charcot-Marie-Tooth (CMT). Until recently, there was little hope for a treatment to help the thousands of children living today with CMT... nor for the thousands more being born each year. That is changing, however, as possible therapeutics are moving rapidly from the laboratory to the clinic!

HNF has launched an on-line pediatric neurology survey to gain a better understanding of your evaluation techniques in assessing young patients with CMT and your perspective of the impact of CMT on their quality of life.

Just a few minutes of your time can strengthen our mission to improve the lives of the many children and families affected by CMT!

YOUR INPUT IS INVALUABLE: WWW.HELPCMTKIDS.ORG

EXCITING CORPORATE NEWS:

Invitae, A Genetic Information Company, Acquires AltaVoice, HNF's Global Registry for Inherited Neuropathies (GRIN) Platform Host

BY **JESSICA ROBERTS**, WRITER, HNF

HNF community partner Invitae recently announced its acquisition of patient-centered data company AltaVoice. Combining the strengths of these two companies enables the creation of new offerings to advance research and provide access to improved care for patients with inherited and rare diseases. As a result of joining forces, the two companies are launching a more extensive data network called the "Invitae Genome Network." This new initiative combines genetic information and clinical data into one extensive, comprehensive network to drive insight into rare diseases.

This development is exciting news for our CMT community, as the "Invitae Genome Network" will ultimately continue to enhance HNF's research goals of connecting patients with health care professionals, scientists, and therapeutic developers to accelerate the understanding, diagnosis, and treatment of hereditary diseases.

Allison Moore, HNF Founder/CEO, expressed: "Participation in clinical trials is essential to develop treatments for rare diseases; however, it is often very difficult for patients to find the right trials and for researchers to find the appropriate patients. Efforts that help match and connect patients to resources are essential in our fight to improve patient care, find cures for CMT, and make the right connections for clinical trial patient recruitment."

As a patient-centered data company since 2007 with a global platform for collecting, curating, coordinating, and delivering safeguarded data from patients and clinicians, AltaVoice (formerly Patient Crossroads) has been developing programs for more than 400 diseases by working with more than 100 advocacy groups, the National Institutes of Health (NIH), Patient-Centered Outcomes Research Institute (PCORI), as well as biotech and pharmaceutical companies. Bringing together Invitae's ongoing testing business with AltaVoice's database of more than 75,000 patients holds the potential for combining valuable capabilities, technology, and data to enhance the use of genetic information for the diagnosis and treatment of hereditary diseases.

Both Invitae and AltaVoice share the common commitment that patients own their data. They believe permission-based sharing of patient data can be valuable to improving patient outcomes and that patients should decide what's best for them. By using technology to remove barriers to diagnosis and treatment. So, patients stay firmly in charge of their genetic information. Patients in the Invitae Genome Network will be able to manage their information based on their genotype and phenotype, allowing them to participate in new research, clinical trials or other treatments that will benefit them.

TO JOIN THE GLOBAL REGISTRY FOR INHERITED NEUROPATHIES:
CLICK HERE

2015 HNF TD Bank
Five Boro Bike Team

TD Five Boro Bike Tour: Meet Team HNF

BY **COURTNEY HOLLETT**, FUNDRAISING COORDINATOR, HNF

The Hereditary Neuropathy Foundation (HNF) is thrilled to have been chosen for the eighth year as a Charity Partner for the 2017 TD Bank Five Boro Bike Tour. This means more participants riding to increase awareness of CMT and thousands more dollars raised to fund research into treatments and a cure!

Held annually on the first Sunday in May, the 2017 TD Five Boro Bike Tour is America's largest cycling event. The event provides participants the unique and fun experience to bike through all five boroughs of New York City—a 40 mile, traffic-free ride for 32,000 cyclists. Starting just north of Battery Park, the tour runs up Manhattan, through Central Park, around a brief loop in the Bronx and down to the Queensboro Bridge passing countless New York City icons on the way. After a ride over the Pulaski bridge passing through Brooklyn, riders enjoy an incredible view from the lower deck of the Verrazano Bridge.

HNF is thankful for the Team CMT members who ride year after year and fundraise for CMT research. Your dedication is making a difference in the lives of all affected by CMT. In addition, we have many NEW members this year including patients, family members and friends. We thank you for you time and efforts as you help us raise funds for treatments and a cure for CMT.

PLEASE CONSIDER DONATING TO A HNF TD BIKE TOUR HNF TEAM MEMBER: CLICK HERE

TO BECOME A TEAM CMT MEMBER: CLICK HERE

Q&A with Dave Washabaugh
HNF Team Member

Q: Tell us about yourself: Where do you live? Favorite hobbies?

A: I'm 53 years old and live in Newark, Delaware with my wonderful wife Nancy. We have three grown sons, none of which show any signs of CMT. I enjoy cycling and ride every chance I get. I love to play golf, but difficulties with my balance have made it a little more challenging. I'm an avid sports enthusiast and a diehard Philadelphia Eagles fan. I love craft beers and brewing my own beer is a passion of mine.

Q: When were you diagnosed with CMT? Briefly explain your journey getting diagnosed with CMT.

A: I was always very active and played football and lacrosse in high school and college. When I was in my mid 30's, I noticed that my feet were getting numb — first my toes and then the balls of my feet.

I really didn't think anything of it until I mentioned to my father that it felt like I was wearing socks all the time. He said his felt the same way. He never complained about it, but I noticed his feet "slapped" the ground when he walked.

I finally went to a neurologist when I was 43 and was given an EMG. The neurologist also ordered a spinal tap to rule out MS and I was eventually diagnosed with Peripheral Neuropathy. The doctor said I could get a blood test to determine what type of PN I had. I declined because the test was very expensive. My feet slapped the ground and my balance wasn't great, but it didn't really slow me down.

Three years ago I got a severe case of the flu and was out of commission for about three weeks. After I recovered, I decided to go out for a run to get in shape for our church's annual 5K. I got about 100 yards from the house and it felt as though I had forgotten how to run. I had no strength in my ankles and my balance was worse than ever. Later that year, I finally had a blood test done and was diagnosed with CMT1B.

Q: Do other members of your family have CMT?

A: My father had CMT, but was never tested. My sister also has CMT and is getting a blood test in March.

Q: Why did you choose to participate in the TD Bank Five Boro Bike Tour?

A: I'm riding because I love to cycle and it's a great opportunity to raise awareness and money for CMT. Only my close friends know that I have CMT or even what is. When I post my donor page on Facebook a lot more people will be made aware of CMT.

Q: What challenges will you face, if any, participating in the TD Bank Five Boro Bike Tour?

A: My biggest challenge is balance and with all the other riders, I get a little nervous.

Q: What are your plans on training for the TD Bank Five Boro Bike Tour?

A: My plans are to ride every chance I get weather permitting. I also plan to keep exercising and stretching.

A good friend of mine, Ian Croft, and I ride every chance we get. We just finished planning out our rides for 2017. We will begin with the Icicle Century ride in March. The TD Bank Five Boro Bike Tour will be our second major ride of the year.

Q: How has CMT changed your life?

A: Although it's slowed my pace down when I walk and affected my balance, it's also affected me in a positive way. I participated in the CMT Summit last October and was able to meet other patients and caregivers from all over the world. Their positive attitudes and courageous struggles have given me such inspiration.

Q: What advice can you give to others that are affected with CMT?

A: Keep exercising and stretching your legs. I think that pushing myself especially when I ride has improved my muscles. I know that we've all probably heard or read that you shouldn't exercise to exhaustion, but as an athlete I don't know how else to exercise. Always push yourself to do more than you think you can.

A: What's a favorite quote of that you think of often?

A: My favorite quote actually comes from a song, My Body, by Young the Giant: "My body tells me no, but I won't quit cuz I want more, cuz I want more." The song plays over and over in my head when I ride, especially heading up a steep hill.

Q: Anything else you would like to share with us?

A: I feel truly blessed. My CMT developed later in life and it is progressing very slowly. Although my neurologist says that I won't need braces or a cane until my mid 60's (about 10 years from now), I'm out to prove her wrong.

Q&A with Rhonda Shumaker

Rhonda is making her mark in dog agility competitions...and she has CMT. We had to know more about how she got involved in a sport that is challenging for anyone, let alone for someone dealing with the daily struggles of living with CMT.

Q: When were you diagnosed with CMT? Briefly explain your journey getting diagnosed with CMT.

A: I was diagnosed with CMT when I was about 8 years old. The disease runs on my father's side of the family. He also had CMT, as did his uncle and his brother. Some of his siblings do not have it. I participated in a study when I was 17 in 1980 that was conducted by the University of Iowa. They took blood samples from all of my family members. That was before they knew about all the different types of CMT. One of my first cousins has recently had the genetic testing done, and she has CMT1A.

When I was growing up, I was always the slowest runner in my class! I have some bad memories about not being able to run fast enough during gym class. As a teenager, I took horseback riding lessons and rode almost every day for five years. I think the strenuous exercise of riding every day may have helped delay the growth of the disease for me at that time.

My symptoms began getting worse when I was in my mid-thirties. I was working a desk job for several years at that point, and had lost a lot of the muscle tone I had when I was younger. When I would go on vacation and do a lot of walking in one day, I would get extremely sore in my hips and knees. I was not using any braces or canes to get around, but I had to use a handrail to go up and down the stairs and could not walk without assistance on uneven or icy surfaces.

When I was 49, my husband and I purchased a blueberry farm in Warwick, RI. He was retiring from his job as a Federal employee at the Naval Undersea Warfare Center, in Newport, RI, but he was only 55 and he needed a new job to keep him busy. I quit my desk job, and we both worked full-time running the farm.

My physical abilities improved drastically in the first two years! There is a hill that we have to walk up that goes from the blueberry field to the house and, at first, I could hardly make it up every day without a hand from Joe. Now, I am able to run up the hill and try to do that at least twice a week in good weather to keep my muscle tone. I can now go up a staircase without a handrail, but still prefer to use one going down, and I feel a little more secure on uneven or icy surfaces.

Q: Do other members of your family have CMT?

A: I have two children, ages 22 and 26, who are not showing any signs of CMT at this point. My two brothers and several of my cousins have CMT.

Q: What is dog agility? How did you start getting involved?

A: Two and a half years ago, we decided to get a new dog. Our dachshund, Fritz, passed away in 2013, and we really missed having a dog around the house. We researched all the breeds

and narrowed it down to a whippet. Looking for a short-haired, floppy-eared, medium-sized, friendly, and house-loving dog. We picked up our new puppy, Viktor Krum, around Thanksgiving in 2014. He is the love of our lives!

His breeder, Phoebe Booth of Shannon Whippets in CT, showed him in conformation classes, and he scooped up a ton of points and got his championship in only six months. He currently is 4 points shy of his Grand Championship, and we are still going to a few conformation shows a year, trying to get those last 4 points. While we were at our first big conformation show in Springfield, MA, we watched what was going on in one of the other buildings for a while, dog agility! I was totally captivated by this amazing sport. Each dog and handler team go out in the ring alone—with the dog off leash—and complete a series of jumps, tunnels, and obstacles. Speed and accuracy win the game. I wanted to do that!

Q: What challenges do you face with CMT and agility?

A: We signed up for our first foundations in agility class when Viktor was 10 months old. Viktor and I had already completed six months of obedience classes, as well as an amazing online course called Recallers by Susan Garrett! The Recallers course gave us the necessary skills we needed to work off-leash in any environment. The agility skills came easily for Viktor, as he is a confident and fearless dog and is up for anything!

The biggest problem we faced in the first six months of agility training was getting me to run. Agility is a fast-moving, high-energy sport for both the dog and the handler, and you have to be able to run to do it successfully. I hadn't done any running for 30 years! My ankles and knees were very weak, and my gait was slow and awkward. But I was determined to keep up with my amazing dog! I started out running tiny, short distances in the house. My ankles, feet, and knees were creaking away as I took my first tentative running steps across the floor.

Q: How has having CMT and participating in agility changed your life?

A: During the time when I was beginning to run again, I found a CMT Facebook page, and that inspired me a lot! There were people running marathons with CMT! I had no idea. Then I found out about the Turbomed FS3000 AFO braces that a lot of the runners were wearing. I ordered a pair for myself online. I started wearing them to agility classes and loved them! The braces gave my ankles more stability and my feet and legs more lift. My balance improved drastically. I felt more confident to go faster without fear of injury. I began running longer distances at home, eventually working up to a mile without stopping.

I still struggle with the longer distances because I don't have the stamina, but what I need to compete in agility is to be able to sprint and do complex turns while running for about 60 seconds at a time. It is a lot to ask of my legs some days. I need to do some sustained fast movement every day, or I find my legs will get that buckling feeling towards the end of my handling runs. In the winter, when I can not run outside, I train for 15-30 minutes a day on a stationary bike.

Q: What are your long term goals for agility?

A: After some very low moments, when I felt I would never be fast or agile enough to keep up with my whippet in the ring, we are now doing an amazing job! We got our second AKC Novice agility title in February of 2017. We are entered in several trials each month this year, and hope to keep moving up the ladder, getting our Q's, or qualifying runs, at each trial. Viktor and I attend group classes twice a week, and I am taking two agility courses online called Handling-360 and Agility Nation, also by the amazing Susan Garrett. My local agility instructor, Beth Szczygiel, is so supportive and encouraging. She does not let me make excuses or claim that I am unable to do any of the handling moves. She teaches a very athletic handling system called One Mind Dog that complements the more distance-oriented handling system of H-360.

So far, I have gone injury free. I have fallen a couple of times, feet and or dog getting in the way, but other than feeling a bit shaken up, there has been nothing to worry about. If I have a sore knee or sore foot, I have found that it will go away in a few days if I keep moving. Hot soaks in the tub with Epsom salts help too. But I still get out there and move, even if there is some soreness. I occasionally take an ibuprofen before class or a trial if I am feeling stiff or sore. Since we purchased the farm, I have lost about 15 pounds, which is a huge benefit to being able to move faster. I now weigh 135 pounds and am 5' 7" tall. I could stand to lose another 10 pounds. One of my favorite quotes that I think of when I am feeling heavy is "to lose weight, you just have to move more and eat less." It is a lot easier to do that in the summertime.

One of the best things about the sport of dog agility is that everyone who participates is so supportive and accepting of each other. It is mostly women over the age of forty-five. We all have our individual pluses and minuses, but when we are in the ring we are all judged on merit alone. It is not a beauty contest. There are no politics or prejudices involved in the scoring. You win if you are the fastest with the least number of penalties. So everyone is on the same playing field regardless of her experience or connections to the dog world. While watching others run their courses, it is my chance to learn. What is a good handling move for each team and what isn't? Can I try something another handler did successfully, with my own dog? Someday I hope Viktor will be among in the top ten whippets in the country in agility. This ranking is based on the yards per second the dog covers while running a Masters level course successfully. It is a lofty goal, but one that I think we can achieve. Look how far we have come already!

Q: What advice can you give to others that are affected with CMT?

A: My advice to fellow CMT athletes and aspiring CMT athletes is to never give up! You can always improve and you are only competing against yourself! Keep a journal to record your progress. It will amaze you to read about what you were doing just a few years ago and inspire you to see how far you have come.

Find a mentor—like my own, Susan Garrett—from whom you can learn. Conquer your fear of failure. No one is judging you, so just do your best and enjoy the thrill of trying. People will be so supportive as they know how much courage it takes to enter a competition.

There is nothing in the world that is better for me than running an agility course with my dog. It is an exhilarating rush that is so addicting. Even if I have problems or make huge mistakes, I love trying and doing the best I can.

Disease Awareness Problem

BY **COURTNEY HOLLETT**, FUNDRAISING COORDINATOR, HNF

If you walked up to a group of people in your town, your city, your school, or your neighborhood…and ask them if they have ever heard about Multiple Sclerosis, the majority would say, "Yes."

Now, ask them if they ever heard of Charcot-Marie-Tooth...the majority would say, "What?"

Even though the incidence of CMT is greater than that of MS and other diseases, CMT goes largely unrecognized—and not just among the general population. Patients go years without a proper diagnosis and care because many health care professionals (HCPs) are not aware of the disease or how to manage it.

Why are we still battling a disease awareness problem? This critical issue was addressed in the closing keynote at the 2016 Patient-Centered Charcot-Marie-Tooth Summit. Robert Moore, husband to Allison Moore, CEO and Founder of HNF, led a compelling discussion on the challenges of CMT awareness and how we—as a community of patients, researchers, and HCPs—can give this disease the recognition it deserves.

He encouraged us to keep pushing the boundaries of raising awareness within the CMT patient and HCP communities. Without awareness, we don't have a voice.

You won't want to miss this video of Robert, who not only educated but entertained the audience with his engaging keynote.

CHECK OUT THE VIDEO ON OUR WEBSITE:
CLICK HERE

Thank You Iris Adler For Your Dedication!

BY **COURTNEY HOLLETT**, FUNDRAISING COORDINATOR, HNF

On January 27, 2017, Iris Adler held her 8th H.E.L.P. Fund Card Party Brunch and Boutique at the Broken Sound Country Club in Boca Raton, Florida. This year, the event set a record attendance number at 192, and it was a tremendous success.

This year was bittersweet for HNF, as Iris is stepping down from the HNF board and focusing her efforts on the Hydrocephalus Association. Iris' granddaughter was born with this rare disease and, like CMT, it has no cure.

HNF is forever grateful for ALL that Iris has done for the CMT community with her fundraising efforts. She will be greatly missed as she is a true team player! We thank her for contributing her time and talents and we wish her much success.

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 for the program is to ensure that access to care results in positive outcomes for each individual patient's clinical experience. We are honored to have these premier Centers and their leading experts in partnership with us to improve the future for people with inherited neuropathies.

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UPCOMING 2017 EVENTS

Save the date!

5/7/17
TD Five Boro Bike Tour
New York City

6/17/17
Sunset Cruise
New York City

9/9/17
Scavenger Hunt
Victor, New York

SET SAIL FOR CMT

BY **COURTNEY HOLLETT**, FUNDRAISING COORDINATOR, HNF

On June 17, 2017, HNF will be kicking off our inaugural Manhattan sunset cruise for a night filled with food, drinks, music and exclusive auction items. Enjoy three levels of dancing, full-service bars, lounges, floor to ceiling windows and walk-around decks all while helping patients and families affected by CMT.

All proceeds with go towards the Therapeutic Research in Accelerated Discovery (TRIAD) program, a collaborative effort with academia, government and industry, to develop treatments for CMT.

FOR MORE INFORMATION PLEASE VISIT: CLICK HERE

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