HEREDITARY NEUROPATHY FOUNDATION

1-855-HELPCMT
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CMTupdate™

Fall 2014

Collaboration is the Key to HNF Success

We at HNF are so proud of the accomplishments of those scientists we fund and are asking you to please continue to support our efforts. Our Therapeutic Research In Accelerated Discovery (TRIAD) program is a proven collaborative model in the drug discovery process. Through partnerships with leaders in academia, government and industry, we will be part of the first Phase 3 trial from Pharnext, an advanced clinical stage biopharmaceutical company for the most common form of CMT, CMT1A.

We are also committed to our continued strategy for developing treatments for CMT2A. For example, our recent funding to evaluate compounds in the zebrafish model is a giant step toward drug discovery. We are also embarking on other forms of hereditary neuropathies to ultimately accomplish the same goals of getting treatments to patients as soon as possible. Our strategy is to put donors' dollars to work funding the scientists who are doing translational research which will move their ideas to treatments that can ultimately be tested in the clinic. This is not an overnight process, which some organizations would like you to believe; we have to be prepared to invest for the long-haul. It usually takes well over a decade to go from a discovery in the laboratory to something that can go to the FDA for their approval. There are no guarantees. If a scientist finds a promising compound in a screen, the odds are stacked against that molecule ever becoming a drug that a patient takes, and it takes millions of dollars to bring a drug to market. Although HNF is a relatively small organization, we do know this disease better than most drug companies, allowing us to make astute investments. With the help of our Scientific Advisory Board and industry advisors, we look to fund projects that are important and may have been missed by others. You will notice we have a diversified portfolio of projects in different stages in the same way that many of you likely diversify your investments.

In 2014 alone we have announced major collaborations with Pharnext and the Biopontis Alliance for Rare Diseases. These partnerships will help develop a pipeline to bring early and late stage treatments to clinical trials for CMT1A. We have also funded Dr. Andy Grierson working on the CMT2A zebrafish model and Dr. Lucia Notterpek working on microRNA for CMT1A. As always we continue to push forward, and following our Scientific Advisory Board meeting in mid-November, we will have new goals to pursue as well. We will report on these in the next issue.

As the holiday season approaches it is usually a time to reflect on the past year and to position HNF for the years ahead. Our goal to fund key projects to bring treatments to the patient is a marathon, not a sprint,

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.

Inside This Issue

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.

and we need your continued support to see us to that finish line! http://weblink.donorperfect.com/HNFDONATE

Positioning HNF to Translate Treatments to Clinical Trials

In June 2013, HNF made the important decision to refine the organization's mission. We now place more importance on new initiatives that take our research discoveries and translate them to the next phase in the drug discovery process.

We divide our research initiatives into 3 areas:

1. Basic Research: This research covers work from many academics around the globe to government agencies like the CDC.

2. Translational Research: The BioPontis Alliance are helping us transition these early stage assets into potential clinical trial candidates.

3. Clinical Research: We are working with Pharnext and Quest Diagnostics to support clinical research trials and ensure early and accurate diagnosis, supported by the Global Registry for Inherited Neuropathies (GRIN).

We would like to do so much more. We have identified two inherited neuropathies (CMT6 and Autosomal Dominant Optic Atrophy) that result in blindness at a young age and we need more funding to enable scientists to develop screens and fill the gaps in knowledge.

It is impossible to predict the future accurately, but in 2015 we hope to see the first phase III trial for a drug for CMT1A, which will be a major milestone for the CMT1A community. We have worked very hard over the last year to support Pharnext and will continue to do so. We have a lot of work to do to ensure the clinical trials are successful in terms of confirmed patient diagnosis and GRIN recruitment.

HNF has not stopped working for you - the patients, caregivers and advocates - and we pledge that we will not give up our fight to find viable treatments for this disease. The scientists we fund are wonderful people who are working collaboratively to ensure that one day you will get the treatments that will help you and your family. When we talk about CMT, we remind everyone that CMT is truly a family disease, even a friend's disease. It affects everyone in your life. HNF takes this effort to heart, and know we have to continue to work hard to support scientific progress. But be assured, there is finally light at the end of the tunnel!

BioPontis Aliance for Rare Disease

Hereditary Neuropathy Foundation (HNF) is pleased to announce the creation of a joint venture to develop drug candidates for the treatment of Charcot-Marie-Tooth (CMT) Disease with BioPontis Alliance for Rare Disease. http://biopontisalliance.org/ HNF has supported basic research into the cause of CMT and early translational research to develop critical tools for drug development, with the goal that research would lead quickly to treatment. Progress can become stalled because drug development requires a complex array of more engineering like technical capabilities, capabilities not available to patients and academic researchers alike. Also new attention must be paid to patenting and aligning to the interest/requirements of the drug industry players who will be needed to bring treatments to patients.

To ensure success of potential candidates that are ready to enter clinical trials—not at the earlier stage where potential treatments are first being developed and tested in cells or animal models multiple steps need to be taken—BioPontis Rare Disease Alliance can provide the scientific, business and patenting structure to help bridge to those pharmaceutical companies.

In the Joint Venture, BioPontis Alliance will manage the project's scientific execution, regulatory strategy, patenting and business partnering. HNF will continue to manage the other important factors that support the drug discovery process, such as basic and early translational research, build and maintain the Global Registry for Inherited Neuropathies and conduct market research and clinical outcome measure research.

To learn more and/or support the joint venture, please contact allison@hnf-cure.org

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Hot Off the Press

Prevention

One of the ways that some CMT patients first become aware of their disease is when they are given a drug treatment for another disease. This is termed chemotherapy-induced neurotoxicity. Drugs such as paclitaxel and the vinca alkaloids that are widely used in cancer treatment cause severe peripheral neuropathy and in some patients this exacerbates CMT, revealing it perhaps for the first time. Researchers from the Johns Hopkins School of Medicine (http://www.ncbi.nlm.nih.gov/pubmed/?term=ethoxyquin+prevents) have published on how a synthetic antioxidant called ethoxyquin which was approved by the FDA 50 years ago, appears to protect against neurotoxicity in both cells and in animal studies. This treatment did not appear to impact the chemotherapy and also seemed to modulate a protein called heat shock protein 90. This work paves the way for further studies of the neuroprotective ability of this compound and possibly clinical trials for patients with pre-existing CMT that need to undergo chemotherapy and for which there are few options. Ethoxyquin as a method of preventing neurotoxicity could spark interest in the search for other drugs that act similarly.

Diagnosis

Accurate diagnosis of CMT is important if we are to identify patients for future clinical trials with treatments for the disease. Currently a tiered approach to genetic testing is used and recommended by clinicians and relies on nerve conduction velocity assessment, disease inheritance pattern and population frequency. The previously recommended multi-tiered decision algorithm is laborious. A recent study by Quest Diagnostics and Baylor College of Medicine (http://onlinelibrary.wiley.com/doi/10.1002/mgg3.106/pdf) has evaluated over 17,000 patients using a variety of gene testing methods. The scale of this study is at least 10 times larger than previous analyses. It showed that 78.6 of those tested were positive for copy number variations of PMP22. The genes GJB1, MPZ and MFN2 were present in 6.7,

RESEARCH PROGRESS - THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY

DRUG DISCOVERY

Discovery Biology

- CMT 1A assay development
- CMT1A/1E high content screens
- CMT 2A mouse model
- CMT 2A assay development
- CMT2A low content screens in zebra fish

Translational Medicine

- “Proof of Concept” studies on an in vivo mouse model with CMT 2A
- Transgenic CMT 1A rats for testing various therapeutics
- Partnering with pharma/biotech (in process)
- “Proof of Concept” for RNA and CMT1A

DRUG DEVELOPMENT

Clinical Trials

- Partnering with pharma/biotech (in process)
- New outcome measure/biomarker for CMT1A/CMT2A (grant submitted)
- Alternate outcome measures for CMT (grant submitted)

Patient Outcomes

- Global Registry for Inherited Neuropathies
- Diagnosis Patient education (Quest/ Athena Diagnostics)
- Quality of Life study (grant submitted)

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5.3% and 4.3%, respectively. So nearly 95% of the patients had mutations in just 4 genes. This represents an opportunity for changing the algorithm for CMT diagnosis such that initially it focuses on testing just these 4 genes.

Treatment

Two recent papers provide some encouraging news in the quest for treatments for CMT1A. A group at the Max Planck Institute of Experimental Medicine in Gottingen, Germany recently described how the CMT1A rat model (in early postnatal development) could be treated with a recombinant human growth factor called neuregulin-1 (http://www.nature.com/nm/journal/v20/n9/full/nm.3664.html). This appears to activate a signaling pathway and is able to improve the differentiation of Schwann cells in CMT1A. However it was found to be less effective in treating older animals. It does open the door for using compounds that modify signaling pathways and the kinases involved. A second paper by a group at the NIH (http://pubs.acs.org/doi/abs/10.1021/cb5005492) used genome editing to create an assay for high throughput screening to expand the targets for drug discovery in CMT1A. The result of this work was the identification of the protein kinase C modulator bryostatin which lowers PMP22 expression. Interestingly this compound was not identified in previous screens by the group which had delivered the proteasome inhibitors such as Borteozomib. In summary, independently two groups have focused in on the role of kinases in pathways that control PMP22. This may open the door for a broader consideration of the many compounds already available that modulate different kinases.

Visit www.hnf-cure.org

Thanks for Participating

HNF wants to give a shout out to all our followers that participated in The University of North Carolina Greensboro Survey. The goal of the study was to learn more about the experiences of people with Charcot-Marie-Tooth (CMT) with genetic counseling and genetic testing. The hope of the survey is to help researchers to better understand what people with CMT think about genetic counseling and genetic testing, and to learn more about their experiences in receiving genetic services. We are looking forward to what Ms. Elizabeth Francisco found from the countless CMT'ers that participated. Thank you for making a difference in the lives of all that are affected with Charcot-Marie-Tooth by giving us 10-15 minutes of your time.

Glossary of Terms for the Drug Discovery Process

Basic research: Highly speculative research that could be pursued for a very long time without any potential application. Sometimes called "blue sky research". Occasionally such research results in great advances to the field that are awarded Nobel prizes. Rarely does this lead to drugs.

Applied research: Research that can have a practical impact in a relatively short period of time. Such research is designed to deliver results that are more likely to yield treatments or new approaches to therapy.

Valley of death: The process of translating basic research into a viable product. The NIH define it as the period of transition when a developing technology is seen as promising, but is too new to validate its commercial potential and unable to attract the necessary funding for its continued development.

Pre-clinical: Any research before a clinical trial. So this includes discovery which encompasses high throughput screening, target validation, lead optimization, toxicology and process chemistry.

Clinical trials: Usually at least 3 phases (Phase I, II and III) before a data package that can be submitted to the FDA for new drug application approval.

GLOBAL REGISTRY FOR INHERITED NEUROPATHIES

Support Hereditary Neuropathy Research by joining the Global Registry for Inherited Neuropathies (GRIN)

Dear CMT Update Readers,

If you or anyone you know has a form of inherited neuropathy, please take 10 to 15 minutes to complete your GRIN registration and upload copies of genetic results and NCV reports, if these have been performed. Impacted adults can complete their own registrations. If you have two or more impacted children, please create an entry for EACH impacted child. Please also create an entry for deceased patients.

It is incredibly important that genetic counselors also forward this email to inherited neuropathy patients. Each form of inherited neuropathy is rare, and most forms are ultra-rare (less than 600 Americans). Unless we begin collecting information on patients, biotech companies will not invest in therapy development for such rare conditions without data to show that they will have an audience for their drug, and possible participants for clinical trials. ONLY de-identified data will be shared with both researchers and companies. i.e. No patient names nor other identifiers will be shared. Academic scientists will receive requested data at no charge. Industry will be charged a small fee to help sustain the annual fees associated with the registry which HNF is paying.

The Global Registry for Inherited Neuropathy (GRIN) is co-funded by the Hereditary Neuropathy Foundation and Hannah's Hope Fund for GAN. I think you will notice a great deal of time was spent developing the questionnaire. It is exciting that we will have the ability to approach a biotech focused on pain pathways and be able to tell them, "We have 3,000 patients with CMT1A and of those, 2,600 have unmanaged pain; and of those, 2,000 said they would be willing to consider volunteering for experimental trials". Or, "We have 150 patients with CMT2E and of those, 140 said their symptoms are severe and rapidly progressing; and of those 100 said they would be willing to donate specimens to help advance research".

Please share with everyone you know who may be impacted by inherited neuropathy, or who have an undiagnosed neuropathy that isn't auto-immune related.

Together, we will make a difference.

Visit www.neuropathyreg.org

Pain and Charcot-Marie-Tooth —Is There a Role for Magnesium?

LORI SAMES

Occasionally you will hear someone say that X helps disease Y and "you should try it". X could be something as simple as a supplement and Y could represent a rare disease like Charcot-Marie-Tooth (CMT). We cannot stress strongly enough that it is important not take anything without first consulting a healthcare professional. CMT is no exception, regardless of how apparently mundane that proposed treatment may be it is not worth the risk.

Here at HNF, we have had recent exchanges of emails with two patients who were magnesium deficient. According to their description, their lives changed dramatically after adding magnesium supplements to their diets. I decided I needed to explore the science on what was known with a broad inherited neuropathy audience.

First, I searched PubMed to see if there were any scientific studies that had been conducted specifically on the effects of magnesium on CMT. Unfortunately no papers were found. I then searched to see if a correlation had been made with CMT and magnesium deficiency. Again, no such correlation has been published as far as I can tell. Clearly it makes you wonder whether anyone has explored the role of magnesium for CMT and just not published it. If there are any scientists reading this that can shed any light please let us know.

Magnesium has however been tested for neuropathic pain and the authors described *"that magnesium diminished the frequency of pain paroxysms and improved the emotional component of behavior in patients suffering from neuropathic pain." But they also suggested "the clinical trial displayed a large placebo response and could not demonstrate any significant difference in pain alleviation after a month of oral treatment between magnesium and placebo".*
http://www.ncbi.nlm.nih.gov/pubmed/21659058

In simple terms magnesium did not work for neuropathic pain. If you have CMT or any form of inherited neuropathy and had any mineral deficiencies that required treatment, that in turn impacted your underlying disease, please let us know by sending an email to: coordinator@neuropathyreg.org.

GET INVOLVED

New Faces of HNF

HNF continues to grow with adding four key members to the team. We are thrilled to have them and look forward to working together to find treatments and a cure for Charcot-Marie-Tooth.

DONNA CUSIMANO, Treasurer

Donna Cusimano, is a real estate attorney whose practice focuses on real estate and complex commercial lending transactions. Donna earned her JD and Masters of Law in commercial real estate from The John Marshall Law School located in Chicago, Illinois in the spring of 1999. After graduation, Donna returned to New York City where she spent 3 years with the law firm Feldman & Markman, LLP and 12 years with the firm Todtman, Nachamie, Spizz & Johns, P.C. Donna represented small community banks and small corporations secure financing for property acquisitions. Donna has recently relocated to south Florida where she intends on continuing to practice law. She looks forward to helping HNF in its mission to raise awareness and find a cure for CMT.

DEBI HOULIARES, Board member

Debi Houliares is the Director of Programming at the EquiCenter, Inc. a non-for profit therapeutic riding facility as well as Head Riding Instructor. Debi holds certifications as an international Therapeutic Riding Instructor and Equine Specialist in Mental Health and Learning. Debi joined the HNF board to raise funds for CMT Type 6 and spread awareness for her son Zachery, who was diagnosed with CMT Type 6 in January 2014. As a side note she believes that therapeutic riding would benefit folks with CMT too! Debi resides in Victor, New York and is married with three children.

KERIN REILLY, Board Member

Kerin Reilly is currently the Director of Admissions for The American Academy of Dramatic Arts, New York campus. She is responsible for the management of the New York Admissions Department. Aspects of this position include recruitment, outreach, admissions, informational presentations and handling VIP visitors. In addition to working on the Admissions side she also taught Public Speaking for the Adult Accelerated Degree Program in both the hybrid online format and traditionally at the Mount Saint Mary College's military satellite campuses which are located at West Point and Stewart Air Force Base. Kerin joined the HNF board to bring awareness to CMT. Her beautiful, brave daughter, Dakota has CMT and Kerin plans to do everything in her capacity to help her daughter and all those living with CMT. Kerin resides in New York and is married with two children.

BERNADETTE SCARDUZIO, Social Media Coordinator

Bernadette (Berns) comes to HNF with a wealth of knowledge about Charcot-Marie-Tooth (CMT). She plans to assist HNF with social media and interact daily with patients and family members that are affected by CMT. She is a Certified Personal Trainer and received her certification from the National Personal Training Institute (NPTI) but had to halt training due to CMT. She also worked for her family business Cuz N' Company Salon and Spa that her dad started 30 years ago and took on many roles throughout the years. In addition she worked in the community as a teacher's aide for Family Support Services. Berns is the star of the "Bernadette Documentary" which follows her journey with CMT. She is social media guru and is looking forward to continue the upward growth of HNF's social media. Bern resides in Drexel Hill, PA and enjoys spending time with her family and dogs.

A Successful September

KERIN REILLY

HNF is proud to share the news of another successful CMT Awareness Month. The HNF community worked hard this past September to build awareness, raise funds for research, and educate as many people as possible about Charcot-Marie-Tooth (CMT) Disease and the impact it has on those living with it everyday. Although we had a serious mission, much of our efforts were rooted in fun activities that spread not only awareness but also a lot of smiles! Thirty days were spent hosting local and national events, launching fundraisers, and spreading the word about the affects of CMT on patients and families.

CMT has been referred to as "the biggest disease you've never heard of." HNF wants to change that. Approximately one in every 2,500 people (2.6 million worldwide) live with CMT; alarmingly, symptoms are often not diagnosed properly and are routinely overlooked by doctors and medical professionals.

The goal this past September was to expand our community through sharing our passion for helping people further understand the seriousness of CMT.

At the start of the month, we launched the Un-selfie4CMT Text-2-Give Campaign. This campaign was very successful and proved to be a fun and engaging event. We raised over $4,600 which means that over 400 people participated by uploading their photo and texting a donation of $10. Short videos to build awareness for this campaign were filmed in NYC early in the month and were viewed on YouTube over 28,000 times and generated hundreds of new likes on Facebook.

During the week of Sept 15th HNF partnered with California Pizza Kitchen for a fundraising event in which the restaurant chain donated 20% of the all checks that were accompanied with a special coupon directly to HNF. Participating locations included NYC, Philadelphia, Sacramento, Seattle, Glendale, and Virginia. The NYC Park Avenue location is on board for next year, after perceiving this event as one of their most successful fundraising events to date.

On September 22nd we held the second annual Dick Sharpe Memorial Golf Event at the prestigious North Hempstead Country Club in Port Washington, New York. All of the proceeds - over $65,000 - went to support our research through the Therapeutic Research in Accelerated Discovery (TRIAD) program.

A Spin for a Cure event on September 27th was held at CRANK NYC. Supporters put on their spin shoes and cycled for a cure! A delicious brunch followed at Parlor Steakhouse where members of HNF and CMT patients and family members mingled and made connections that we believe will lead to future collaborations. This fun event raised $12,000 that will go to support HNF's TRIAD research program.

The HNF 'Inspire' online support community welcomed 68 new members this September showing us that the word is spreading. HNF is extremely happy that so many people participated in the September CMT Awareness events. We look forward to next year!

EVENTS

Grace's Courage Crusade

Grace's Courage Crusade, a passionate campaign of the Sidoti/Caldarone family to raise awareness and fund CMT research, held its annual "Brunch by the Beach" fundraiser in scenic Newport, RI on November 2, 2014. Now in its seventh year, this event has become a beloved tradition for the crowd of over 150 attendees, and participants enjoyed great food, fun games, exciting raffles, and of course, fantastic desserts. (Who says brunch doesn't come with dessert?!)

This year was the most amazing ever, with over $15,000 raised for the Hereditary Neuropathy Foundation's TRIAD research program. Sean Ekins, HNF's Chief Science Officer, presented a CMT research update during the event, outlining the exciting developments in CMT research, including the upcoming Stage III clinical trial for CMT1A, ongoing CMT2A research using a zebrafish model, and progress on the development of a CMT2A mouse model. (See past HNF newsletters for all the latest research developments and sign up to receive these important newsletters in the future.)

We at HNF, and the Sidoti and Caldarone families, would like to thank all of the generous supporters who have joined with Grace's Courage Crusade and HNF to make a real difference for kids like Grace who live with the challenges of CMT every day.

Walked the Red Carpet!

A Hollywood Affair drew dozens of people dressed to the nines to walk the red carpet at the Wingate, Wyndham Hotel in Lexington, South Carolina. Food and drinks were available, and guests were entertained by music from DJ Curtis Wilson. There was a great silent auction featuring vacations, gift certificates for local businesses and signed items from celebrities including Taylor Swift, Jadeveon Clowney and Tony Romo.

All the proceeds from this event ($3800) will go towards Jillian's Cure to fund research for an autosomal dominant optic atrophy. The disease causes the nerve endings in a person's eyes and ears to die, resulting in blindness and deafness.

Jillian's mom, Carolyn Nava, is working to raise money to go toward research for a cure, hoping one can be found in time for Jillian. She is already at work planning another event, the Jillian's Cure Super Hero 5k, set for April 18, 2015.

For more information visit www.carolynnava.com

Meet Team CMT U.K. Member, Julie Glover

The Hereditary Neuropathy Foundation and Team CMT have a global reach. We have Team CMT members in Canada, Australia, Finland, Turkey, Iran, Vietnam and Scotland. Our Scottish member, Julie Glover, recently represented Team CMT at a 5k run in Holyrood Park in Edinburgh Scotland. She did the race with her American friend Shondra. Shondra is a stroke survivor so she also had something to prove that day.

Julie is originally from Rockford, Illinois and is mom to two boys, Caleb (4 yrs.) and Carter (17 months). She's been in love with Scotland since she was a little girl and fell in love with her husband Chris, a "burly Scotsman" as she describes him. She and Chris were married in 2009.

Although she loves Scotland and her life there, she really misses her family back here in the States. She misses her Nani (grandmother), her mom and seeing all of her nieces and nephews grow up. Family is a big priority for her. One of her favorite activities to take walks with her family or just ride the bus together.

Like most of us, CMT is a family affair for Julie. She shares CMT with her grandmother, uncle, and brother. She started to experience leg weakening around age 25 and six months later could not stand on her toes. The doctor told her she had an aggressive form of CMT and would probably be in a wheelchair in 10 years.

So far no wheelchair yet. Julie is a full time student in addition to being a trained fitness instructor and Zumba coach. She is also qualified in KCR (Kinetic Chain Release). She got certified after discovering the treatment and seeing a massive improvement in her legs and quality of life. She became certified to help others in another way.

Helping others is the reason Julie joined Team CMT. She wanted to help make a difference by doing something active and to be an inspiration. She wants to show others that although CMT is progressive, there is no need to give up. She found us from a facebook posting in the CMT U.K. Group.

Julie is passing on that desire to help others to her kids. She has taught tolerance for people with disabilities. One day she was on the bus with her children and her son Caleb saw a young man with a cane board the bus. The bus was very full and Caleb stood up and said to the man "Have my seat. You can rest your legs."

Julie views her experience with CMT as a blessing. Her CMT is not as severe as others, but every day is a challenge to find the energy she needs to get everything done. Sometimes she gets blue, but reminds herself of all the things she can do and everything she has overcome. She feels she would not be the person she is today if she had been born without CMT. The determination and will that comes from being disabled…. especially when someone tells her she can't do something. She laughs when she thinks about how she has proved the doctors wrong.

Running races may seem like a small thing, but Team CMT members continue to inspire others in the CMT community to be more active. We are showing the CMT community that while we have CMT it does not have us. The positive example set by Julie and other team members is so important for those with a condition where there is no treatment and no cure.

Thank you Julie for sharing your story and being such a wonderful role model for your family and those of us in your CMT family!

Team CMT Founder Headed to World Championship

Team CMT founder Chris Wodke is headed to the ITU World Championship. She will be representing Team USA at the World Aquathon Championship on September 16th, 2015. The race is only 90 miles from her Milwaukee home and Chris is no stranger to the course, having raced it twice in the last two years at the Chicago Triathlon. It will almost feel like a hometown race!

The USAT sanctioned sport consists of a 1500m open water swim and a 10k run. Chris qualified at the National Championship in El Reno, Oklahoma on October 5th where she finished 11th in her age group. The top 20 in each age group are eligible for the World Championship.

She is also eligible to compete in the ITU Duathlon World Championship in Adelaide, Australia in October of 2015. She finished first in the PC Open Division at the National Championship race in St. Paul, Minnesota on July 19th. She is going to try to classify as a para-duathlete and if she does not pass classification will race as an age group athlete.

Wodke competed in four national championships this year. In addition to the El Reno and St Paul race, she participated in the Age Group National Sprint Championship in Milwaukee and the Paratriathlon PC Open race in Tempe, Arizona.

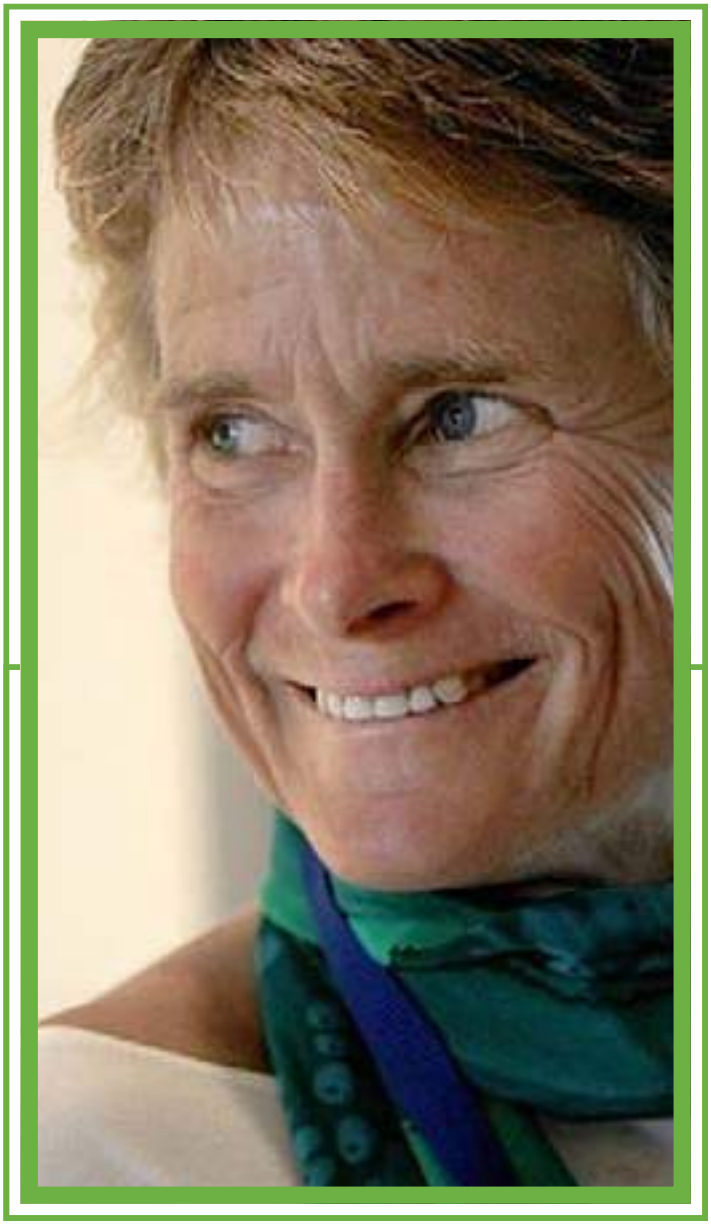
This is not her first time competing for Team USA. She raced for the team at PATCO Dallas in May of 2014 as a para-triathlete where she finished 4th in her category.

It is easy to race as an age group athlete at a national championship. For most of the races there is no qualifying time. Wodke hopes to see other Team CMT members at these events. It would be great to have multiple Team CMT members at a World Championship. Although she knows she cannot be competitive against other age group athletes, preparing for races keeps her motivated to pushing foward…and it's a great way to raise awareness!

A Rare Disease Patient That Does It All!

A movie script could not play out like this. There are very few occasions when a rare disease patient becomes the topic of a story that is truly uplifting. Often the stories are tragic or disheartening. That was not the case, however, with CMT patient Kim Goodsell.

Kim recently became the subject of a story by the acclaimed science writer Ed Yong that simultaneously appeared in National Geographic online (http://phenomena.nationalgeographic.com/2014/08/19/how-an-extreme-athlete-uncovered-her-own-genetic-flaw/) as well as the Welcome Trust blog, Mosaic (http://mosaicscience.com/story/diy-diagnosis-how-extreme-athlete-uncovered-her-genetic-flaw). The article describes how Kim was a competitive triathlete until she was diagnosed with arrhythmogenic right ventricular cardiomyopathy (ARVC), predisposing her to sudden cardiac death. In the years that followed Kim's motor skills became progressively compromised and thirteen years after her ARVC diagnosis, she was diagnosed with CMT 2B1, co-segregating with an Emery Driefuss Muscular Dystrophy (EDMD)-like myopathy. As unbelievable as it sounds, Kim had multiple rare diseases.



So Kim set out to learn as much as possible to understand whether her diseases were related. She eventually narrowed down the culprit to a gene called LMNA. She then had to convince scientists to dig deeper and went as far as funding the sequencing of the DNA of her LMNA gene. The results showed that Kim indeed had a mutation in LMNA. Kim ultimately wrote up a thesis on her whole experience and is trying to get her experience published. This unique ability to not only survive, but thrive, and drive research to diagnose the culprit for her own diseases has attracted interest from prominent scientists for several reasons. For one it shows how much a patient can do themselves when empowered to do so. And second, as a patient Kim knew more about her disease than anyone else including the specialists that looked into her diseases. CMT (and rare diseases in general) need patients like Kim that can make discoveries that could lead to a greater understanding of the disease. This inspirational story is not over and it will be fascinating to see what Kim does next on her voyage of rare disease discovery.

Thank you, Kim, for sharing your story with HNF…you are an inspiration!

A Profound Act of Kindness

ALLISON MOORE

Every so often, a seemingly random act of kindness can change everything. Some would call it a miracle, a blessing, a gift from the universe. HNF was the beneficiary of such an act this month, when a very kind-hearted person left us a legacy to help those affected by CMT, a disease that had an impact on our benefactors own life. When I arrived at the HNF offices last week, my first task, as always, was to open the mail. Although it is not my favorite way to start the day, it is just one of those tasks that you have to get done. (continue on pg. 13)

Kara Q&A

My name is Courtney Hollett, Fundraising Coordinator at the Hereditary Neuropathy Foundation. This time of year I count my blessing daily and wanted to share with you a Q&A session I had with a new supporter of HNF. Kara, like myself has many family members affected with CMT and I reached out to her to share her thoughts and advice about how to "support" a friend or family member that's affected with Charcot-Marie-Tooth.

COURTNEY: **Tell us about yourself Where you live? Favorite hobbies?**

KARA: I grew up in Crystal Lake, Illinois and lived in Chicago for 5 years before moving to New York City. I currently live in Manhattan.

I love to travel internationally and spend time with my family in the Chicago area--my Mom, Dad, two sisters, two brothers-in-law, and my favorite little munchkins -- my niece Kenzie (4 1/2 years old) and nephew Jack (2 years old). I also enjoy photography, animals, and the great food, wine, and culture of NYC, including Broadway musicals and plays.

COURTNEY: **How are you affected by CMT?**

KARA: My Grandmother, Mother, and sister have CMT.

COURTNEY: **Why are you fundraising for CMT?**

KARA: I'm fundraising to raise awareness and support CMT research efforts.

It is no surprise to me that CMT is often referred to as "the biggest disease you've never heard of." Growing up, I always thought my sister had just inherited my Mom's feet, muscular build, and coordination. My family had never heard of CMT. For years, my Mom and sister frequently sought treatment from doctors and podiatrists. It wasn't until about 10 years ago that a podiatrist on staff told my sister about CMT during an office visit to an orthopedist for a knee injury. The information available on the disease at that time was limited. Even now, my family has seen several doctors who have never heard of the disease.

Hopefully raising awareness will help doctors diagnose the disease early, help to educate those impacted regarding different treatment options, ways to manage the disease on a daily basis (including understanding those medications that are contraindicated for those with CMT), as well as provide hope that current research will lead to a treatment for CMT.

COURTNEY: **How has CMT changed your life?**

KARA: CMT has significantly impacted my entire family.

Most recently, my sister underwent several major foot/ankle surgeries. It has been very difficult, both physically and emotionally, yet my mom and my sister continue to amaze me. My sister works each day to improve her strength and recover with two young children at home. Of course, my Mom and the rest of my family are right by her side.

After connecting with HNF and learning more information about CMT, I am more determined than ever to raise CMT awareness and contribute to research efforts to find a treatment or cure.

COURTNEY: **If you care for someone with CMT do you have any information for others?**

KARA: Be supportive and informed. Try to learn about the disease and don't be afraid to talk about it. While it may be a difficult and emotional subject to discuss initially, if you care for someone with CMT, I believe that being open about it helps show your support and helps you understand and be sensitive to your loved ones' day-to-day challenges.

For years, I have felt both upset that my mom and sister have CMT - and completely helpless. Becoming involved in fundraising efforts and helping to raise awareness has been the most effective and rewarding way for me to show my love and support. It has helped me to feel empowered and feel that I am doing something to make a difference in their lives and in the lives of others impacted by CMT.

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

KARA: "Our lives are not determined by what happens to us but how we react to what happens, not by what life brings us but the attitude we bring to life."
—Wade Boggs

Boot Season is Here!

Fashion tips for those wearing leg braces…

DAKOTA REILLY

Leg braces can be extremely helpful for those of us living with CMT. Braces can make a world of difference in our ability to walk longer distances, improve balance and coordination and maintain energy.

Of course, I would never recommend hiding the fact that you have CMT but if sometimes you feel like covering your braces for the sake of fashion, here are some fall style tips……

Doesn't it seem impossible to find an outfit that works with leg braces?! Yoga pants with sneakers is a common go-to for me however, I absolutely love to dress up so I've been experimenting with different boots. For example…

This photo is from my high school pep rally; I usually don't wear a tutu - ha! I usually tend to wear boots with leg warmers! Traditional leg warmers however can sometimes be too loose and do not stay put. Because of this I purchased knee high socks and then cut off the feet so that they can fit over my braces as leg warmers. I find that this works better than actual leg warmers because the socks are tight and stay up above the brace throughout the day. Boots are very hard to find, you just have to get lucky. I find that boots with a sturdy, wide bottom, and have a full zipper on the side work best.

Here is another example…

Boots that have laces like these are very convenient to put on over braces and provide room for your feet! I found these boots for only $30 at Target! They come in black, burgundy and floral print. I simply put the knee socks over the braces and then added traditional loose leg warmers over that. An extra layer of socks over the brace will smooth the rough edges of the brace that show through your pants/ socks.

When the weather is hot and I don't feel like wearing leg warmers, I will wear loose pants like this!

Any of the shoes you currently own can work with this look, for me, my low-rise boots are the most comfortable. The pants

in this photo became a trend recently which is a great option in the warmer months! You can wear a crazy pattern like this or a solid color; they are sold at many stores so they are easy to find! What is really great is that they are as comfortable as yoga pants but can be made to look fancy if you are in the mood to dress up! Boot cut jeans also work for this look.

If I had to sum up what works best for me when dealing with leg braces, I would say leg warmers, boots (especially ones with laces), sneakers and loose pants.

I hope my tips are helpful and have provided you with some new and interesting ideas!

For more information, feel free to check out the HNF Pinterest page and look for upcoming tips from myself and Estela.

HEREDITARY NEUROPATHY FOUNDATION
www.hnf-cure.org
432 Park Avenue South, 4th Fl.
New York, NY 10016
@CMTNeuropathy
HereditaryNeuropathyFoundation

A Profound Act of Kindness

(continued from pg. 10) This day, however, the mail would hold an amazing but bittersweet surprise. A bulky envelope containing many documents informed me of the passing of a HNF supporter and fellow patient. In the cover letter, it was stated that HNF was to be the beneficiary for this individual's considerable IRA. Reading about this generous gift brought tears to my eyes. It made me reflect on how short life is and her strong belief in our mission and our patient first philosophy. It is our intention to use this bequest to further impact our TRIAD research initiatives by funding projects that will lead to treatments for CMT.

How Your CMT Legacy Can Live On

Through effective planned giving, you can balance your personal financial goals, realize significant tax benefits and help your relatives and fellow patients that may be afflicted with CMT and/or related inherited neuropathies by bequeathing all or a portion of your estate to the Hereditary Neuropathy Foundation.

Some of the ways to give include IRA's, annuities, life insurance and trusts.

For more information on planned giving, please contact Donna Cusimano at Donna@hnf-cure.org or simply call 212-722-8396.

Upcoming Events - Save the Date

H.E.L.P. Brunch, Boutique & Card Party
Friday, January 23, 2015
Boca Raton, Florida

Super Hero 5K Jillian's Cure
Saturday, April 18, 2015
Lexington, South Carolina

TD Bank Five Boro Bike Tour
Sunday, May 3, 2015
New York, New York