HEREDITARY NEUROPATHY FOUNDATION

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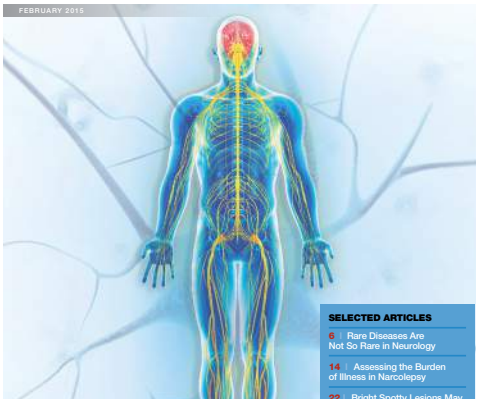
NEUROLOGICAL RARE DISEASE
SPECIAL REPORT




# CMTupdate™

Winter 2015

## Breaking News: First Therapeutic Gene Therapy to Treat an Inherited Neuropathy is Approved for Clinical Trial!

The first disease community to receive a therapeutic gene to the spinal cord for an ultra rare inherited neuropathy is Giant Axonal Neuropathy (GAN). Congratulations to Hannah's Hope Fund (HHF), a 501(c)3 public charity, which has driven this collaborative research in less than six years. Six million dollars has been raised to date to fund pre-clinical and clinical research on this rare disease.

The Phase 1 trial is recruiting - info here: **Intrathecal Administration of scAAV9/JeT-GAN for the Treatment of Giant Axonal Neuropathy** patients. A benign viral vector known as adeno associated virus serotype 9 (AAV9) is the "Fed-Ex truck" delivering a healthy copy of the GAN gene to the nerves in the spinal cord of affected patients. It is administered by a lumbar puncture to the cerebral spinal fluid. This trial will be housed at the National Institutes of Health, in Bethesda, MD.

If the AAV9 vector is well tolerated in the central nervous system, this gene therapy vector can be used for any genetic disorder whose targets are motor and sensory neurons, and whose causal gene size is small enough to fit inside the viral capsid. This is a turning point for rare neurological diseases and the inherited neuropathy community in particular. The impact of this work can potentially help other forms of Charcot-Marie-Tooth (CMT) and related diseases. Our hope and prayers for a successful outcome go out to all the families with GAN!

## Finding Experts Health Care Providers for Charcot-Marie-Tooth

HNF has a new initiative aimed at identifying expert Health Care Providers (HCP) for the CMT community. We are creating an innovative **HCP Directory** which provides these clinicians with additional tools to care for their patients. We will provide them with the best standard of practice guidelines, as well as verifying them as a provider to ensure quality diagnosis and treatment to the CMT community.

Once endorsed by HNF, we will provide these HCP's with a special logo to add to their website, a link to take a free continuing medical education course and a special Essential Guide Booklet with all they need to know about CMT.

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



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.

A secondary goal of creating the HCP Directory is to encourage their patients to join **Global Registry for Inherited Neuropathies** (GRIN) so they can collaborate in the research process by accelerating therapy development for the CMT community.

To launch this exciting initiative, HNF recently participated in the **Neurological Rare Disease Special Report** that was published as a supplement to **Neurology Reviews** and now available as a digital version under the "Education" tab.

HNF developed a two-paged advertorial describing the diagnosis and treatment for CMT (p. 8-9). The Neurological Rare Disease Special Report was mailed to over 24,000 clinicians, including 20,000 neurologists and 4,000 primary care physicians in the U.S. In addition, this special report will also be distributed from the Neurology Reviews exhibit booth at over 15 major industry conferences. Our goal is to continue to raise disease awareness, inform clinicians of how to recognize CMT, accurately diagnose and finally inform them of future potential treatment options. We will continue to highlight CMT through similar mechanisms and peer reviewed publications.

## Scientific Advisory Board Meeting

**SEAN EKINS, CSO, HNF**

On the 7th of November we convened our scientific advisory board meeting at the HNF offices in NY. Our meeting included **Renée JG Arnold, PharmD, RPh** (President & CEO, Arnold Consultancy & Technology LLC), **Robert Burgess, PhD** (Faculty of The Jackson Laboratory in Bar Harbor, Maine), **Joel Freundlich, PhD** (Associate Professor of Pharmacology & Physiology and Medicine at Rutgers University–New Jersey Medical School), **Steven J. Gray, PhD** (Research Assistant Professor Dept. of Ophthalmology, U. of N. Carolina at Chapel Hill), **Joseph J Higgins, MD, FAAN** (Director of Neurology Quest Diagnostics, Athena Brand, Marlborough, MA), **Brett Langley, PhD** (Director of Neural Epigenetics at the Burke Medical Research Institute, Assistant Professor of Neurology and Neuroscience at Weill Medical College of Cornell University), **Jun Li, MD, PhD** (Associate Professor, Department of Neurology, Director of Charcot-Marie-Tooth Clinic, Faculty Member in Vanderbilt Brain Institute and Faculty Member in the Center for Human Genetics Research at the School of Medicine, Vanderbilt University.) **Nadia Litterman, PhD** (Collaborations Director at Collaborative Drug Discovery (CDD)), **Lucia Notterpek, PhD** (William T. and Janice M. Neely Professor and Chair, Department of Neuroscience at the University of Florida, Gainesville), **David Pleasure, MD** (Professor of Neurology and Pediatrics, Director of Research, Shriner's Hospital for Children, Director, Institute for Pediatric Regenerative Medicine and Distinguished Professor, UC Davis), **Michael Sereda, MD, PhD** (Professor of Neurology and Group leader in the Department of Neurogenetics, Max Planck Institute (MPI) of Experimental Medicine, Göttingen, Germany), and **Dianna E. Willis, PhD** (Head of the Laboratory for Axonal and RNA Biology, Director of the Center for Pain Research at the Burke Medical Research Institute and an Assistant Professor of Neuroscience at Weill Cornell Medical College). In addition **Barbara Handelin, PhD** from the Biopontis Alliance was in attendance.

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After a welcome from Allison Moore (CEO, HNF), I presented a brief overview of the work HNF had funded to date and our goals. Each scientist gave an overview of their latest research related to CMT or related areas of study, whether it was funded by HNF or not, and described the challenges and opportunities. For example Dr. Sereda updated us on his recent work that described how Neuregulin 1 looked promising for reversing CMT1A. Dr. Notterpek described her miRNA project for CMT1A. Dr Li presented how MRI technology was used to see changes in the peripheral nerves which correlates with CMT and hence may be used as a measure of the disease. Dr Pleasure updated us on the CMT2A mouse characterization. Dr. Willis and Dr. Langley described their research which covered the MFN2 mouse and HDAC6 inhibitors, respectively. Dr. Burgess works with various mouse models and focused on characterizing the CMT2D mouse. Dr. Gray outlined his work on gene therapy, including the research on gigaxonin for Giant Axonal Neuropathy (which should be going to clinical trial in the next few weeks). Dr. Higgins summarized his recent publication which analyzed the frequency of gene mutations in 17,377 patients with CMT and showed that just 4 genes can be used to capture most patients with CMT. Dr. Litterman summarized published work on the use of stem cells and how collaboration could have an important role in making the research process more efficient. Dr. Freundlich described how as researchers are generating high throughput screening data, they could be learning from it with computer models and at the same time understanding what molecules should be avoided. Dr. Arnold stressed how patient reported outcomes were instrumental in getting drugs approved as companies increasingly have to show an increase in quality of life to justify the cost of treatment. This led us very nicely into a discussion of what we should be funding in the future and where the gaps are that will impact upcoming clinical trials. We have written a **detailed review** that has been published and captures all of the discussion and make this freely available to the scientific community.

## Support CMT Therapeutic Alliance

BioPontis ALLIANCE FOR RARE DISEASES

Hereditary Neuropathy Foundation has spent the past seven years and over 1.3 million funding basic to early translational research and now the time has come to move these discoveries towards the goal to provide treatment options for patients. HNF has entered into a joint venture – the CMT Therapeutic Alliance - with a unique non profit organization (BioPontis Alliance for Rare Diseases) that brings professional drug discovery capabilities to translate our research results into potential treatments. The CMT Therapeutic Alliance is supporting a project with SYSTASY Bioscience GmbH (located in Munich, Germany) to screen drug compounds in a novel cellular model of CMT1A. The scientific advisor of the study, Prof. Michael Sereda from the University Medical Center Göttingen, Germany, says: 'We have recently seen that the demyelinating pathology in CMT1A appears to be the result of an imbalance between two chemical pathways in Schwann cells. We will be looking for drug compounds that can correct this imbalance. If candidate compounds can be validated in pre-clinical animal models, then clinical trials are in immediate reach.'

In order to facilitate the first phase of this initiative, we have set a fundraising target of $500,000.

To learn more, **click here.**

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## Hot Off the Press – Potential Treatment for CMT1A

Two recent publications from Pharnext describe a novel synergistic combination of 3 drugs (baclofen, naltrexone and sorbitol) and its effect on CMT1A both **in the lab** and in a **phase II clinical trial**. These 3 drugs already approved but for unrelated conditions, are combined at new optimal lower doses and under a new formulation. This novel potential therapeutic is called PXT-3003.

In preclinical studies, PXT-3003 was shown in vitro to synergistically increase myelination of axons co-cultured with CMT1A rat Schwann cells. The combination of drugs was also shown to lower PMP22 expression (which at high levels in CMT1A is responsible for dysmyelination and consequently axonal loss and muscle atrophy). In the rat model of CMT1A different measures of effectiveness suggested that PXT-3003 was also promising and likely efficacious. The very low doses of all three components would also negate any adverse side effects.

The phase II clinical trial used three dose levels of PXT-3003 in 80 adults with mild to moderate CMT1A. The trial confirmed the safety of the combination drug and the best improvement was seen at the highest dose. PXT-3003 was safe and well tolerated. PXT-3003 showed, beyond stabilization, a significant improvement in the Overall Neuropathy Limitation Scale (ONLS) versus the placebo group. ONLS is a major scale to evaluate disability of upper and lower limbs for peripheral neuropathy. This represents the most promising potential treatment to date after ascorbic acid (vitamin C) failed to demonstrate efficacy for CMT1A in various clinical trials.

There are still gaps in understanding the mechanism of how PXT-3003 actually exerts its effect. It is hoped that by looking at patients over a longer period and possibly treating them earlier before the disease clinically affects them, this may improve nerve conduction and potentially halt and reverse disease. PXT-3003 represents a new hope for CMT1A patients in years, but there is still a long way to go (several years) before it may be more widely available as an FDA and EMA approved treatment for CMT1A.

An International Phase 3 trial will enroll later this year both in the US and Europe.

If you are interested in participating in the clinical trial, **join** the Global Registry for Inherited Neuropathies (GRIN).

## Do You Have Charcot-Marie-Tooth Type 4 (CMT4)? – We Need Your Help

Autosomal recessive demyelinating CMT4 patients present with early-onset and slowly progressing symptoms. These include progressive distally accentuated weakness and atrophy of muscles in the lower limbs. In addition, patients may have weakness and atrophy of hands, sensory loss and pes cavus (high-arched feet), and walking difficulties. Further information on CMT4 can be found at **Orphanet**.

Many genes and their mutations are associated with CMT4 including GDAP1, MTMR2, SBF2, SH3TC2, NDRG1, EGR2, PRX, FGD4 and FIG4. These have different roles, for example GDAP1; the most frequent genetic cause of CMT4 is a protein on the outer mitochondrial membrane and regulates the mitochondrial network. MTMR2 encodes a protein that may have a role in neural membrane recycling and membrane trafficking. A more recently identified gene mutation in **SURF1** encodes cytochrome c oxidase, a protein anchored to the mitochondrial inner membrane. Some of these genes like MTMR2 and FIG4 interact to control the phospholipid substrate PtdIns (3,5)P2 in neurons and Schwann cells to **control phospholipid metabolism**. Clearly understanding the complexity of mutations in

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one or more of these genes and predicting the types of symptoms observed is difficult as these genes regulate proteins with a multitude of functions. There are still gaps in understanding this disease. To date there is also no treatment for CMT4. The prevalence is also unknown. We would like to evaluate potential early stage therapeutic treatments for CMT4 and you can help! Please register if you have CMT4 at **GRIN** and this may give us an estimate of prevalence and you can find out more.

## Glossary of Terms for Gene Therapy

**Gene Therapy**: The transplantation of a normal gene into cells in place of missing or defective gene in order to correct genetic diseases.

**Giant Axonal Neuropathy (GAN):** A single gene, axonal neuropathy. Most cases of GAN are severe, and fatal in the early 20's. However, there are slower progressing patients with GAN.

**Intrathecal Administration**: A route of administration by injecting into the lumbar space, delivering drug products to the cerebral spinal fluid.

**Lumbar Puncture**: Spinal tap.

**AAV9 Vector:** Adeno Associated Viral Vector Serotype 9.

**Viral Vector**: A benign virus that acts like a Fed-Ex drug, delivery of healthy genes to cells.

**Viral Capsid:** The part of the virus that contains a healthy gene.

## 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 (Pharnext)
- 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)
- Clinical Disease Awareness (Neurology Reviews/NORD)

GLOBAL REGISTRY FOR INHERITED NEUROPATHIES

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

## Now GRIN is Accessible in 141 Different Languages!

**BY LORI SAMES**

The **Global Registry for Inherited Neuropathies (GRIN)** has implemented Google Translate. Why did we choose Google? Because Google is leading the way in teaching computers how to interpret meaning, avoiding the traditional method of decoding language.

Software that decodes language can turn 'kindergarten' into 'children garden'. Instead, "Google mines existing translated material, recognizes how words or phrases typically correspond, and uses probability to deliver the best match based on context".[1]

Having the ability to communicate with patients in 141 different languages will hopefully result in more patients joining **GRIN** globally to prepare for future clinical trials, for example, Pharnext's PXT-3003 phase III clinical trials for later this year.

[1]Ref: **https://translate.google.com/**

## Accelerating Patient Access to Investigational Drugs in 2015

**BY ALLISON MOORE**

Currently the FDA is working to update the process for physicians applying for accelerated patient access to investigational drugs, while the drug or biological product is being tested in clinical trials. This will also be important for the CMT community as clinical trials for this disease are being launched. This is termed "expanded access" whereby there is no other product that can diagnose, monitor, or treat the patient's disease or condition, and the patient is not and cannot be enrolled in a clinical study for various reasons (such as patient inclusion criteria or access to a clinical site).

"Compassionate use" was coined early in the HIV/AIDS epidemic when the FDA authorized unapproved investigational drugs for patients in certain cases. The application process was then complex and time consuming. It could take up to 100 hours for physicians to complete the form. FDA recognizes physicians demanding schedules and is simplifying the process to a shorter form for completion within 45 minutes.

The most recent 2015 drafted process is entitled **"Individual Patient Expanded Access Applications: Form FDA 3926,"** it includes a simplified application form that, when finalized, will be used for requesting the medications, and is designed to greatly simplify and accelerate the process by which a physician can request that FDA permit the use of an experimental — so-called "investigational" — drug or biological product while it's still being tested to establish its safety.

The bottom line for patients is that while a clinical trial is in process they can apply to access the drug for their disease.

For more information:

**http://blogs.fda.gov/fdavoice/index.php/2015/02/a-big-step-to-help-the-patients-most-in-need/#.dpuf**

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## Taking Matters Into Your Own Hands

Do you have a rare form of CMT?

We were contacted recently from a patient with CMT2D who was looking for advice on how to make a difference. Our recommendations for all people with an ultra-rare form of CMT who want to get involved:

1) **Join** the global registries for inherited neuropathies (GRIN).

2) Join the Inspire Community at: **Charcot-Marie-Tooth (CMT) Support Community - Inspire.**

3) Create a Facebook page specifically for CMT2D or your form of CMT and encourage all patients that find you to join the **GRIN**.

4) Go **www.pubmed.com** and do a search on CMT2D or your form of CMT and to learn where your form of CMT is being worked on. Reach out to the authors of research papers (an email address should be included at the bottom of the abstract).

5) Email every scientist in the world working on your form of CMT and ask if they need a skin biopsy from patients to advance research. Hopefully, your local university has success in growing out skin fibroblast cell lines following punch biopsies. They would grow out a large number of skin fibroblast cells that they would freeze into 10 separate vials. You could then have a vial shipped to 10 different labs studying your form of CMT. They would ship overnight, on dry ice. *HNF can help with this process if cells are needed by researchers.*

Initially, the most important thing is to find other patients with your form of CMT and ask if they too will have skin biopsies performed. If you are able to fundraise, you can earmark the funds for your type of CMT, through the Hereditary Neuropathy Foundation. We can help identify what the status of the field is for your form of CMT, and help prioritize research initiatives that need funding.

With over 80 different CMT causal genes now identified, patients must become active and helping to drive research. Except for CMT1A, CMT2A and CMTX, all other forms are considered ultra-rare, meaning less than 600 Americans are impacted.

Rare forms of CMT must unite in order to help fund, and drive research, to advance to the point where a pharmaceutical company may be interested in further investing in an ultra-rare CMT. Seed funding is needed for basic research until a discovery is made that peeks the interest of industry. TOGETHER, we will make a difference for ALL forms of CMT!

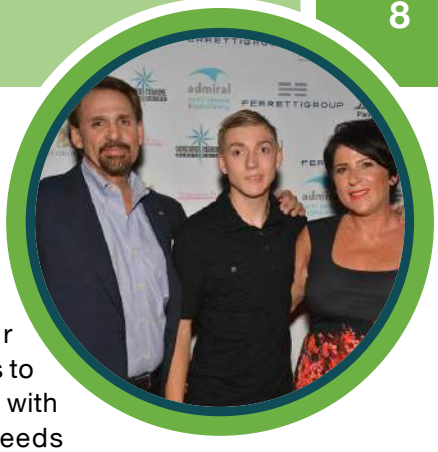
## WHAT IT'S LIKE TO LIVE WITH CMT

Visit our website for your free copy of our latest Essential Guide, What It Is Like to Live with CMT. Based on the findings of the renowned Ph.D., Elizabeth Barrett and Dr. Carol Birdsall, the booklet provides a moving and informative account of the daily challenges and triumphs experienced by people with CMT.

# GET INVOLVED

## The Houliares Family Takes on CMT6

**BY DEBI HOULIARES**



We have two choices in this world. We can sit back and do nothing, allowing this disease to happen, or we can go out and fight to make a difference.

When Zach was first diagnosed with CMT6 after 14 years of doctors trying to figure it out, we were fighting mad that there was a disease out there that no one has ever heard of. How can this be? After researching on the internet, which didn't provide a lot of information, my first call was to the HNF, and I spoke with Allison Moore. She was the very first person that was able to answer so many questions about CMT that all the doctors could not.

Zach's form of CMT is different, though. He is legally blind, with a vision of 20/400. With a rare form of a rare disease, our new mission in life became raising awareness and money for research for Zach and others like him. There could be many other people who are not even diagnosed because doctors are not aware of what CMT6 is. So we are taking matters into our own hands and spreading the word.

Zach loves to play sports but can't physically play on any school teams due to his blindness and difficulty walking. But one week after this diagnosis, Zach was unexpectedly sent onto the basketball court in a game with a minute left. Unscripted and unrehearsed, his teammate threw him the ball and swish, all net. There wasn't a dry eye in the house. The **video** played on the local news and even ESPN.

That was only the beginning. We started raising awareness in Zach's schools by selling "Find a Cure for CMT" bracelets. Most local business where we live in Victor, NY have a jean day where if employees pay $1, they can wear jeans to work with proceeds going to HNF "Team Zach." Within 10 months of Zach's diagnosis we had a successful fundraising event at my husband's workplace and raised quite a bit of money. We already have a motorcycle/vehicle scavenger hunt event planned for July of this year and a second event is in the works. People are willing to support a good cause, and it's our job to make these events happen, no matter how big or small. Everyone sees and know the symbols like the pink ribbons for breast cancer, puzzle piece for autism. Inherited neuropathies are just as big and as powerful. Fighting to raise awareness for CMT has to start somewhere, so why can't it start with us. Never give up hope. To learn **more**.

## Double Your Donation

Corporate employee matching gift programs: What are they and how do they benefit HNF's Therapeutic Research In Accelerated Research (TRIAD) program?

Corporate matching gift programs are charitable giving programs setup by corporations in which the company matches donations made by employees to eligible nonprofit organizations. For example, if a donor works for Bank of America and donates $100 to the Hereditary Neuropathy Foundation, Bank of America will double the donation by also writing a check for $100.

Thousands of companies offer matching gift programs. A couple examples include:

- Johnson & Johnson – Triples donations with $2:$1 matches for current employees while also doubling donations for retirees.
- Home Depot – Matches donations $1:$1 up to $3,000 annually per employee.

The impact of these programs can be substantial! In 2011, Microsoft matched $48.9 million worth of employee donations to schools and 501(c)3 nonprofits. Fifteen million employees work for companies with matching gift programs.

Are you one of the millions working for a company that offers matching gifts? HNF has set a goal for 2015 to raise $10,000 from these programs. By meeting our goal, $20,000 will go directly to CMT Research.

For more information, visit our **website**.

## New HNF Team Member, Joy Aldrich Inspires Others Through the CMT Inspire Patient Community

Joy joined HNF in February 2015 to focus on the growth of HNF's online patient support community as a CMT Advocacy Director. Joy and her husband, Toby, live in Seattle, WA, with their beloved yellow lab, Charley. While food service distribution was her career; finding a treatment for CMT, traveling and cooking are her passions. You may remember her food blog, **www.apassionateplate.com**.

The diagnosis of CMT came when Joy was a teenager, after years of trips and falls and sprained ankles. Her mom and brother were also diagnosed at that time. Years went by before Joy noticed the more rapid progression of CMT symptoms and pursued a genetic diagnosis, which was confirmed as CMT1A. At that time, she turned to HNF for advice on AFOs and a letter writing campaign to tell family and friends about her diagnosis. Since then, Joy has participated in HNF's CMT Awareness month **video campaign** and hosted fundraisers in the Seattle area.

Joy is excited about promoting, growing, and strengthening the **Inspire CMT support community** as it is a valuable social media tool which connects patients to patients and caregivers in order to share important information about CMT. She invites you to create your profile and join the discussions today!

You can also read Joy's story by **clicking here.**

## Q & A with Andy Heck

Our Q&A for the Winter CMT Update is Andy Heck. Andy has been diagnosed with Charcot-Marie-Tooth since four and doesn't let CMT stop him for living his life to the fullest!

**COURTNEY: Tell us about yourself. Where do you live? What are your favorite hobbies?**

**ANDY:** My name is Andy Heck and I currently live in East Troy, WI. I will soon be relocating to Charlotte, NC for work. I work as a Multimedia Marketing Specialist at Sealed Air. My favorite hobbies are film, bicycling, and cars.

**COURTNEY: When were you diagnosed with CMT? What type of CMT do you have?**

**ANDY:** I was diagnosed with CMT1A when I was two years old.

**COURTNEY: Do other members of your family have CMT?**

**ANDY:** Yes, several people in my family have CMT including 3 aunts, a cousin, my dad and brother.

**COURTNEY: What are the challenges you face with CMT?**

**ANDY:** As it pertains to my hobbies, I have several challenges.

My doctors always tell me that riding a bike can help me a lot as long as it isn't any type of high impact riding. The problem there is that I've ridden BMX bikes for the past 15+ years. I like going to the skate park and pushing myself to learn new lines and new tricks. I broke my ankle about 12 years ago and since then, I've sprained it countless times. A few years ago, I got an AFO for my left leg and that has helped a lot. It immobilizes my ankle, which protects it; however it makes it harder to feel the pedals. The other issue with the AFO is that my leg gets pinched between it and the seat of my bike when I pedal. I solve this issue with duct tape.

I also like working on classic cars. I have a 1967 El Camino that I restored

several years ago and I'm working on a couple other cars currently. I can usually get the job done when it comes to working on cars, but I've noticed that my hands get very sore after using wrenches. It also causes me to lose strength in parts of my hand which makes it very hard to do some of the things after awhile. I just work on things like that in small doses now to help keep my hands functional.

For film, operating the camera becomes difficult. It is hard to get smooth, moving shots when my body doesn't respond as well as others. I have to find different ways of getting the right shot. It hasn't happened in awhile, but I have dropped cameras… considering I drop just about everything on a regular basis. Despite the challenges, I find it to be a very cool ability that I have to capture video and photos of people doing what they love to do. It allows me to stay involved in sports and other activities that I might not have the ability to fully participate in anymore.

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

**ANDY:** I feel like I've lived a pretty normal life. I can typically do all of the things that any other person could. I just know that if I didn't have CMT, I might be able to do those things better. This really bothers me. I always hear people say that you can do anything you want to as long as you put your mind to it. I agree with this in a way, but the fact is that I have to draw the line somewhere. I like pushing myself physically, but I refuse to push to the point where I'll really hurt myself. I am constantly balancing my hobbies with risks and that is something I wish I didn't have to do.

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

**ANDY:** Know your limits but keep pushing yourself. People in general often deal with their own excuses and insecurities that hold them back. Don't use CMT as an excuse. You are capable of more than you think. You may have to do things a little differently and it may take you a little longer, but that doesn't mean it is impossible and it doesn't mean that you won't get the same satisfaction anyone else would get from the same accomplishment.

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

**ANDY:** "Motion creates emotion."

## Team CMT Takes on the Bermuda Triple Challenge

Meet Team CMT member Bill Morgan. Bill and Liz (his wife) recently ran three races back to back in Bermuda for Team CMT.

"Let us run the risk of wearing out, rather than rusting out."

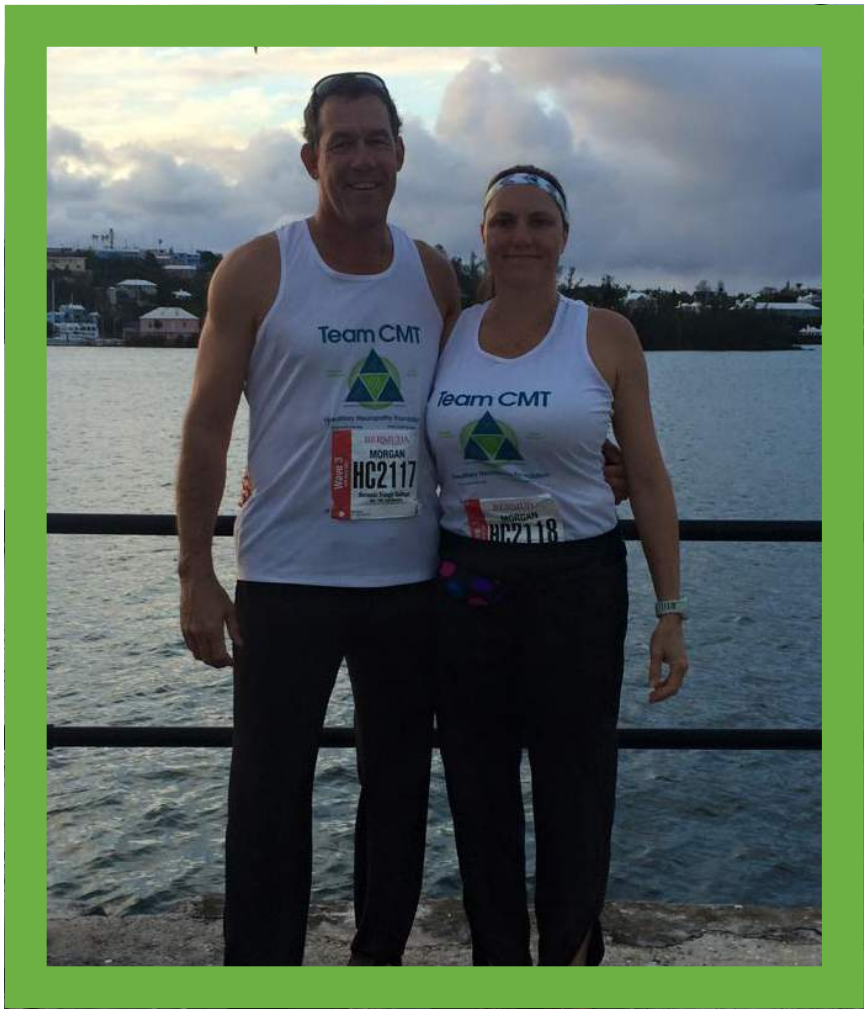
*- Teddy Roosevelt*

Our first race as Team CMT members was the Bermuda Triangle Triple Challenge. It was held in Hamilton, Bermuda on January 16th, 17th and 18th, 2015.

The race consisted of three series, a one mile run on Friday night followed by a 10K on Saturday morning and finishing with a Half Marathon on Sunday morning. The course was absolutely beautiful, but very challenging. Bermuda is very hilly!

The one mile race on Friday night was exciting to participate in as it is the "Bermuda Invitation Mile" and we had the pleasure to see world class runners compete. What a great way to start our visit to Bermuda!

The 10K on Saturday morning took us through the interior of Bermuda and gave us the opportunity to see the landscape of this beautiful island.




We had heard Bermuda was hilly, but we were not expecting what this course held for us. The constant uphill/downhill made this one of the hardest 10K's we have ever run! After finishing and enjoying the rest of the day exploring Bermuda, we prepared for the Half Marathon the next morning.

The Half Marathon on Sunday took us around the perimeter of the island where we were treated to breathtaking views of the beaches and shoreline. This course was also very hilly, but we were prepared for it and had a great race. Liz accomplished a Half Marathon PR!

The people of Bermuda love running and lined all the courses cheering and encouraging all the runners. I can't count the number of times someone yelled "Go Team CMT!" when they saw my Team CMT shirt.

At the finish celebration I had lots of questions from other runners and friends asking what Team CMT is. Most of my friends have known that I have some type of neuropathy, but had never heard of CMT, so having the opportunity to explain what CMT is and how it affects those with it was a good feeling for me.

My CMT is not at a severe stage. I have constant numbness in my feet and toes and regular pain and cramping in my calves. I know that regular stretching and running is the best therapy for my condition. I am fortunate to be able to continue running and I am proud to wear my Team CMT shirt. Hopefully our efforts will help raise awareness and someday a cure so others with CMT can run too.

# Team CMT UPCOMING EVENTS

## Team CMT Updates

Team CMT members are making a difference raising awareness all over the world and funds to support CMT Research. Don't think for a minute that your contribution whether you wear the Team CMT shirt at the gym, physical therapy or participate in an athletic event doesn't have an impact. It certainly does! To date we have 161 Team CMT members, have raised $91,000 to support CMT research and have reached thousands of people worldwide through athletic events, newscasts, and social media.

A special thank you to all our Team CMT members for helping us in our quest to cure CMT. To join Team CMT and make a difference **click here**.

## Bike NY Spotlight

**KRISTIN GELZINS**

Growing up, I was an all star softball player and an avid runner. Nothing could hold me back from just about anything, from skiing to hiking, biking and kayaking. I was in school to get my nursing degree while I worked as an EMT which I loved doing. When I was 21 years old I worked down at the World Trade Centers as an EMT which set my asthma into overdrive. The many attacks I had landed me in the hospital and on steroids numerous times.

At 22 I had lost a significant amount of muscle tone in my legs, and developed avascular necrosis (is the death of bone tissue due to a lack of blood supply). I was sent to a neurologist who at the time treated all of the current problems I was having with the steroid (prednisone). After about a year of seeing no improvement, I wasn't taking any excuses. I was put through test after test. I had both of my hips replaced after failed bone grafts at age 28, and by the time I was 29, I was pregnant and relying on crutches and a wheelchair to get around. I went back to my neurologist and demanded answers. After he was done looking over all of my testing, including the genetic testing that was done years before, the words Charcot-Marie-Tooth came out of his mouth. Now it finally all made sense to me. I now know what I'm dealing with, and I'm glad I pushed for answers. Since I've had my son, I stopped relying so heavily on my wheelchair and started becoming more and more active with him. I play wheelchair softball now, and I tried my hand at modified ice hockey. I still kayak and fish. When it is warm out I would go walking or biking with my son every day. When it's cold we find other ways to stay active. I still have my good days and bad days.

I am signing up for my VERY FIRST Five Boro Bike Tour. I need your help to find a cure for CMT. Please consider **donating** to my fundraising page to help me, my family and everyone living with CMT. Although CMT may have slowed me down a bit, it will never stop me. Thank you so much for your support. It means the world to me!

# EVENTS

## Calling All Cyclists… Enjoy NY Sights While Riding to Cure CMT!

Hereditary Neuropathy Foundation (HNF) is excited to participate for the fourth consecutive year in the TD Bank Five Boro Bike Tour. It's an honor says Allison Moore, Founder/CEO of HNF to once again be chosen as a charity partner in one of the biggest events in New York and the largest cycling event in America.

The event provides participants the unique and fun experience to bike through all five boroughs – a 42-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 Riders will get VIP treatment, which includes breakfast and lunch as well as priority start, special bike parking at the start, and a Team CMT singlet.

100% of all donations will go towards HNF's Therapeutic Research in Accelerated Discovery (TRIAD) program for Charcot-Marie-Tooth. **Click here** for more information and to register.

## Sixth Annual Card Party Brunch

On January 23, 2015 dedicated H.E.L.P. (Help Elliot Live Proud) Fund supporters participated in the 6th Annual Card Party Brunch and Boutique at Broken Sound County Club, Boca Raton, Florida. Record numbers flocked into the picturesque country club for a day filled with gourmet food, raffles, cards and a silent auction! HNF Board member Iris Adler increased this event by 50 new faces this year! New faces equals spreading awareness for Charcot-Marie-Tooth, the number one inherited neuropathy. In addition to card members we had many club members approach us and ask about CMT. One member quoted "what a wonderful event for a great cause, please include me next year". Many members contributed with a donation towards our record breaking total. HNF is proud to report that over $40,000 was raised to fund research for CMT2. The Adler family would like to thank all the participants, volunteers and those individuals that donated that were with us in spirit, but were unable to attend! It's cause of people like you that we WILL find a cure for CMT!

For more information on H.E.L.P. Fund or to support CMT2 research, **click here.**

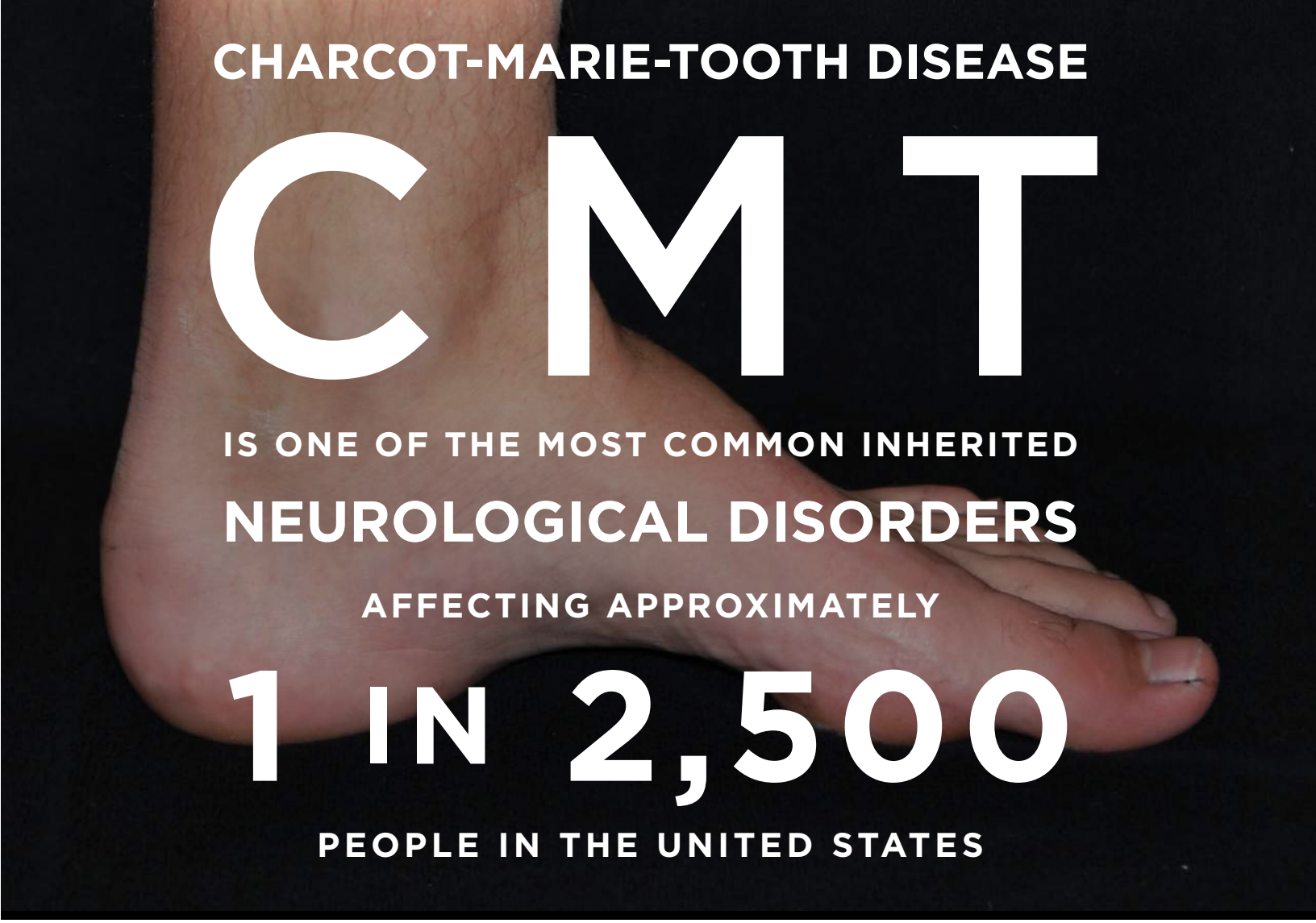
# UPCOMING EVENTS

## Upcoming Events - Save the Date

**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

**The Rochester Chrome Divas**
Saturday, July 25, 2015
Rochester, New York


## Why does it take the average patient 2 years to receive a diagnosis?

Through educational resources and genetic testing, the HNF helps physicians identify, diagnose and treat CMT patients.

Join our provider directory and help us spread awareness and get access to treatment options for CMT patients.

HEREDITARY NEUROPATHY FOUNDATION

Find out more at hnf-cure.org/cmtprovider