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CMT

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CMTupdate™

Spring 2015

New Strategic Alliance with Rare Disease Report (RDR)

By Allison Moore
Founder & CEO – HNF

We are excited to announce our new collaboration with Rare Disease Report (RDR) to increase awareness of Charcot-Marie-Tooth (CMT) among the medical community. RDR's website, e-newsletter, and print publication offers an independent voice for the Rare Disease Community. They strive to bring together medical, scientific, investment, regulatory, and advocate professionals interested in rare diseases and orphan drugs. Since HNF is focused on supporting and developing treatment options for CMT, this collaboration can be groundbreaking towards advancing CMT disease awareness.

Through this new alliance, HNF will expand its outreach to support our new Health Care Provider (HCP) education program, an initiative aimed at identifying expert HCP's to support the CMT Patient Community. This program provides clinicians with additional tools to care for their patients such as best standard of practice guidelines, information on genetic testing, and patient assistance programs. In addition, HNF preferred HCP's gain access to other preferred HNF CMT Providers to support the need for accurate diagnosis, supportive treatments, and other educational materials.

With the help of RDR, we will reach an audience of over 60,000 clinicians through their digital and print distribution channels. Throughout the year, RDR will produce **video interviews** with CMT experts and industry leaders on the diagnosis and management of patients with CMT, as well as provide insight about new research and clinical trials.

James Radke, PhD, Managing Editor of RDR said, "The rare disease community functions best when all parties are part of the conversation. We look forward to working with HNF and provide them with a means to expand their reach into the clinical population." **Click here** to view the HNF-RDR Partnership webpage.

As HNF's Founder & CEO, I firmly believe that it is clear that HCP's are unaware of CMT, how to recognize the early symptoms, and how to manage this and related inherited neuropathies. HNF plans to change that!

Finally, HNF is asking the community to please reach out via email at info@hnf-cure.org and share the HCP's you would like us to be made aware of and why you would like to see them as part of the HCP directory. Please provide name, address, and phone number.

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





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 New Mouse Model for Charcot-Marie-Tooth (CMT2)

By Sean Ekins
Chief Science Officer – HNF

We were recently informed that The Jackson Laboratory (JAX, a nonprofit biomedical research institution headquartered in Bar Harbor, Maine) had taken delivery and will be distributing a newly generated CMT-related mouse model. The new model expresses mutant mitofusin 2, a mitochondrial membrane protein involved in mitochondrial fusion and regulation of vascular smooth muscle cell proliferation.

The creator of the mouse model, Dr. David Pleasure of the University of California at Davis designed the mouse so that researchers have the ability to specifically induce expression of the mutant gene in a tissue of

interest. Dr. Pleasure said, “The heterozygous mouse, which expresses the mutant gene as a knockin in the Rosa locus, shows a mild foot deformity demonstrable by catwalk analysis, as well as an imbalance in muscle fiber types demonstrable by immunohistology and histochemistry. Initial histological analysis of tibial nerve failed to document obvious axon loss. The phenotype in these young adult mice is subtle.” This project was funded by HNF. We look forward to Dr. Pleasure publishing his work in due course. It may require additional work to push the characterization of the mouse further. We have made the mouse model available without license so that other researchers can use and evaluate it freely. We thank The Jackson Laboratory for their considerable assistance and

willingness to distribute this mouse.

Additional information can be obtained at The Jackson Laboratory **strain datasheet**. Orders can be placed for this mouse strain today, and the first mouse should be ready to be shipped by the end of the year.

The JAX mouse repository provides a variety of other mouse models relevant for CMT, and is actively seeking additional models. Please contact Mike Sasner at Mike.Sasner@jax.org.



Glossary of Terms

Axon

Is also known as a nerve fibre, a long, slender projection of a nerve cell, or neuron, that typically conducts electrical impulses away from the neuron's cell body.

Heterozygous

Adiploid organism is heterozygous at a gene locus when its cells contain two different alleles of a gene.

Histochemistry

A branch of histology dealing with the identification of chemical components in cells and tissues.

Histology

A department of anatomy dealing with the minute structure, composition, and function of tissues.

Immunohistology

The microscopic study of tissues with the aid of antibodies that bind to tissue components and reveal their presence.

MFN2

Mitofusin 2 is a gene that encodes a mitochondrial membrane protein that participates in mitochondrial fusion and contributes to the

maintenance and operation of the mitochondrial network.

Mitochondrial

Are a subunit within a cell that plays an important role in generating energy for the cell.

ROSA locus

Is a position used for constitutive, ubiquitous gene expression in mice.

Tibial Nerve

Is a branch of the sciatic nerve and is found in the leg.



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Introducing Our Newest Scientific Advisory Board Member

Dr. Scott Stromatt has a medical degree from the University of Chicago and an MBA from the University of Colorado. He is board certified in Internal Medicine and has over 26 years of biopharmaceutical experience, including three years as a biotechnology analyst for a Wall Street investment firm.

Dr. Stromatt has conducted over 55 Phase 1 to Phase 4 clinical trials in a wide variety of clinical indications, including oncology, rheumatology, pulmonology, gastroenterology, and neurology. In the field of neurology, he

ran clinical trials with a novel growth factor for Lou Gehrig's disease, or amyotrophic lateral sclerosis. He has worked on various regulatory filings including INDs, NDAs, BLAs, and MAAs. Additionally, he has experience in medical affairs with extensive market and brand management support.

Currently, Dr. Stromatt is the Chief Medical Officer at Emergent BioSolutions in Seattle, WA – a biotechnology company focused on oncology, autoimmune disease, and infectious diseases.



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Greensboro Survey Study for Genetic Counseling and Genetic Testing

By Elizabeth Francisco
M.S. Genetic Counseling

The goal of this study was to explore the perceptions and utilization of genetic counseling and testing by people with Charcot-Marie-Tooth (CMT). It's important to understand what people with CMT think about genetic services and what factors may be supporting or suppressing their access to these services. Participants were recruited through the

Hereditary Neuropathy Foundation databases. The project was Internal Review Board (IRB) approved at the University of North Carolina at Greensboro.



THE UNIVERSITY of NORTH CAROLINA
GREENSBORO

Genetic Counseling Results

- 724 participants completed the survey. Not all participants responded to every question, and incomplete surveys were considered for the material that was provided.
- 73% of participants indicated they had never been offered an appointment with a genetic counselor.
- Of those that attended genetic counseling, 86% were happy with their experience.
- Over half of participants who have not had genetic counseling indicated they would be interested in a referral to a genetic counselor.
- 44.6% of participants who had not received genetic counseling perceived cost could be a negative of this service. Only 5.3% of those who did have genetic counseling reported cost as a negative.

Genetic Testing Results

- 53% of participants indicated that they had been offered genetic testing, 7% indicated that testing was not offered because a family member had already undergone testing, and the remainder were not offered testing.
- Of those offered, 79.6% chose to pursue testing.
- 92.5% of those that received testing were happy with their decision.
- Of those who have not had testing, 71.6% indicated they were interested in pursuing genetic testing.
- Those who had genetic testing were significantly more likely to report access to research as a benefit than those who have not had testing.
- Participants who had not had genetic testing were more likely to express concern about potential negatives from this service, including increased worry about themselves and their children post-test, increased chance of discrimination, and most of all cost.
- Only 5% of those who had genetic testing felt that the cost of testing was a negative to their experience. 62% of those who had never received testing anticipated cost would be a negative.



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.

Comments

Many participants who had genetic counseling provided comments about their experience:

“She believed and really listened to my story and was willing to test me (with the geneticist) for the only disorder I had ever come across that matched my symptoms – CMT2C. When the results came back positive, she explained the results in detail and promised to research for more information since it is so rare. Although I am still waiting on her report 3 months later, it was still a very positive experience.”

When asked to elaborate on experience benefits of genetic testing, many participants provided comments:

“[My] Daughter [was] misdiagnosed for 7 years, [it is] nice to [now] know [her diagnosis] and be able to research and help others by being involved with research on CMT1B.”

We also received many comments from participants who are interested in testing, but have not yet had testing:

“I would like to be certain of the type and inheritance of my CMT to counsel my children.”

“My kids need to know if they are carriers of the gene. They’re young adults and planning on having children of their own, but they need to know if they’ll get CMT (adult onset in our family).”

“As part of the Hereditary Neuropathy Foundation’s Health Care Provider (HCP) outreach program, HNF will be working to educate the genetic counselor community on the importance of identifying CMT, proper genetic testing, and providing helpful information for patient cost assistance programs.”

Conclusion

- Genetic counseling is an important and helpful service to patients with CMT.
- Genetic counseling can help people with CMT who are considering testing:
 - Cost was an over-estimated negative by people who had not had genetic counseling or testing. Genetic counselors can research appropriate testing strategies and help people apply for insurance coverage to reduce cost.
 - Participants who have not had genetic testing were also more likely to be worried about the potential for genetic discrimination and increased anxiety after testing. Talking with a genetic counseling may help alleviate some of this concern.
- Meeting with a genetic counselor after testing can help people with CMT understand their results including inheritance, natural history and progression, and research opportunities.
- Barriers to genetic counseling:
 - Although genetic counseling is a great resource for people with CMT, most are not being referred to a genetic counselor.
 - It is important for genetic counselors to reach out to the neurologists in their area to alert them to the services they can offer.
 - Neurologists and other providers should become educated on the benefits of genetic counseling and be prepared to provide people with CMT with accurate information about benefits and potential negatives of genetic counseling and testing, and referrals to these services.
 - Education of the CMT community on what options are available to them and their potential benefits and negatives will allow people with CMT to be their own advocates and request referral to genetic counseling and testing services.



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

HNF Joins The Coalition for Clinical Trials Awareness (CTA)

Awareness of Clinical Trials Week, May 4th – 8th, 2015

By Allison Moore
Founder & CEO – HNF

Whether you've taken a medication today to manage a chronic disease or simply to keep a headache at bay, you know that medical innovations have the power to change lives. But these breakthroughs don't just happen. Developing new medicines is a lengthy and complex process, relying on heavily volunteer participation to evaluate potential therapies for safety and effectiveness. Volunteer participation is also important in pre-clinical work (natural history and outcome measure endpoint studies) leading up to clinical trials. Without the patients who volunteer to participate in clinical trials, the development of new medicines would not be possible.

During Clinical Trials Awareness Week, we recognize the individuals who have made new, innovative treatments a reality for patient's worldwide – clinical trial researchers and participants. By shining a spotlight on these unsung heroes, we can raise awareness about

the importance of participating in a clinical trial.

Clinical trials may benefit participants by providing them the opportunity potentially to receive an investigational drug that is not available to people outside the trial. In addition, they may receive treatment from a clinical research team that provides careful oversight of their medical condition.

For a new medication to receive FDA approval, it must undergo comprehensive testing on actual patients. Yet finding participants for these trials can often be slow, exhausting and ultimately unsuccessful. Research from the Tufts Center for the Study of Drug Development found that eleven percent of trial sites fail to enroll even a single patient. Thirty-seven percent under-enroll, and those who do meet their enrollment targets typically have to double their original timeline.

CTAA is working to educate the public on **what is a clinical trial (see video)**.

We feel strongly that everyone should have access to helpful information about clinical trials and the opportunity to determine if participating in a clinical trial is the right choice for them. We look forward to the day when industry develops clinical trials for the CMT and inherited neuropathy community.

We thank our **Facebook** fans and **Twitter** followers for joining us in supporting CTA Awareness Week.

The Hereditary Neuropathy Foundation is proud to be a member of The Coalition for Clinical Trials Awareness and we look forward to playing a leadership role with this organization and this effort.



Social Media Buzz: Top Facebook Posts #CTAW2015

Clinical Trials Awareness Week was May 4th – 8th and HNF recognizes the unsung heroes – clinical trial researchers and participants who have made new, innovative treatments a reality for patients worldwide. **#CTAW2015**

We hope by this time next year we will be celebrating within our community too. Let's CURE CMT!
45 Shares, 293 Likes

Clinical trials advance scientific knowledge that may lead to innovative treatments. Help us in celebrating Clinical Trials Awareness Week. **#CTAW2015**

Join the Global Registry for Inherited Neuropathies
20 Shares, 137 Likes

Our CMT Inspire Community, Soon to be 2,000 Strong!

By Joy Aldrich
CMT Advocacy Director – HNF

HNF has partnered with Inspire to provide a safe online health and wellness community in which patients, families, friends, and caregivers connect with one another for support

and information. Some of the HOT topics currently being discussed include **“Pregnancy and CMT,”** **“Types of AFOs/Leg Braces,”** and, **“What You Wish Your Doctor Knew.”**

If you have not **joined** us yet, please take a minute to set up your profile

and join our community. Don't forget to add your picture to make things a bit more personal. For more info on the CMT Inspire Community, please contact JoyAldrich@hnf-cure.org

10 Reasons to Join the CMT Inspire Community TODAY!

- 1 Control what you share with extensive privacy settings.
- 2 Get support for your health concerns.
- 3 Join multiple Inspire communities (i.e. Invisible Disease Alliance) to connect with more people.
- 4 Receive regular updates about the health topics you care about.
- 5 Connect with others who are dealing with the same health concerns.
- 6 Learn about treatments and therapies.
- 7 Get real life answers from others that are experiencing your disease firsthand.
- 8 Network with individuals that live in your area for face-to-face support.
- 9 Seek comfort at any hour that you are not alone in your struggles.
- 10 Discover opportunities to participate in clinical trials and other research.

[Click here](#) to join the CMT Inspire Community!

Inspire together we're better™

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.



Accepting Myself for Who I Am

By Laura F.

Growing up, I always knew I was different than my friends. I couldn't run fast, tripped often (the scars on my knees are a reminder), was lousy at any sports-related activity, and was generally weak and uncoordinated. My friends accepted me as I was and didn't really question it, but sometimes strangers would ask, "Why are you limping?" or, "What's wrong with your leg?" I usually just brushed it off with some lame excuse like I was tired, or I "sprained" my ankle. I developed very effective coping mechanisms to hide my differences and, for the most part, it worked well.

Looking back, I think it was more noticeable than I cared to admit, but most people were just too polite to say anything, but when someone was bold enough to ask, the question created a ton of embarrassment for me. I felt ashamed and humiliated. It was a completely irrational way to feel for something I was born with, but because I grew up not knowing there was a disease causing my problems or see myself as "abnormal" I did not even want to acknowledge it.

Charcot-Marie-Tooth (CMT) is an insidious disease that creeps up and

progresses slowly. Gradually, you forget what walking properly actually feels like, or that you should have the ability to move your toes or do squats. So, for the first time in my life, I visited a chiropodist (deals with assessment, diagnosis, and treatment of the lower limbs), who assessed me walking barefoot. The look of shock and surprise on his face was alarming. He asked me to sit down and questioned why no one in my family had noticed the way I walked. How had I managed to go this long without a diagnosis? He used words like "neurological," "serious," and "disease." I was scared and felt like I had just received a death sentence. He confirmed that I had CMT1A. Mystery solved. I was relieved in a way. I didn't care to know about all the things that could happen to me in the future. I just wanted to live my life. I continued on through law school, got married, was focused on work and family. I was in my mid-twenties at that point. Unfortunately, shortly after getting married, the disease seemed to escalate its downward progression.

We wanted to start a family right away. I learned that CMT was one of the diseases that could be tested for using Preimplantation Genetic Diagnosis (PGD). This involves in vitro fertilization process with the extra step of performing genetic testing on the embryos prior to implantation in order to transfer the embryos that are free of CMT. I felt very fortunate that this process was available because it gave me a sense of control over ensuring that my CMT did not persist into the next generation. I was consumed with fear over how my husband would be affected if my CMT was passed on to our child. Would he blame me? Would he resent me? And how would I myself deal with the guilt if my child was more severely afflicted than I was? It was for all these reasons that we opted for PGD. As it turned out, the in vitro process was unsuccessful for us and it was heartbreaking because we knew that all the embryos were healthy. So we let it go and I became pregnant

naturally about a year later. We were prepared for whatever outcome God chose for us and were blessed with a healthy baby girl, followed by another precious daughter a few years later. Many couples are successful with PGD and this procedure may be a viable option for those who are struggling with the decision of whether to have children.

Fast forward 15 years later, two beautiful children, now 40 years old. I am beginning a new chapter in my life. I have let go of the shame. I have let go of the embarrassment. I am telling people that I have CMT. I was fearful about what people would say, what they would think, how I would be regarded. Those self-limiting beliefs that had kept me immobilized and isolated were all in my head. No one cared that I had CMT! Nothing changed in how they treated me. In fact, it has been liberating. It has been a tremendous relief because I do not have to pretend or put up a front anymore! I can just be me. My friends and family have embraced me without making the CMT front and center. It's just a part of me, but not the essence of me.

Now I am doing things that I never would have done, even just a few years ago. Recent changes in my personal life have forced me to wake up and break free of old programs, find the real me, be who I really am, love myself, gain back the confidence that I once had. I look in the mirror and I like what I see and who I am. I am truly grateful for my life and the experiences that have brought me to where I stand today. I have met so many inspiring and amazing people since reaching out to the CMT community. My story is not much different than many others with CMT. It's a journey for all of us. It has ups and downs like anything in life. But I feel CMT places us all on a unique journey, one of compassion, empathy, courage, resilience, and determination. I am so blessed and honored to be a part of the CMT family!



The Long Road to Diagnosis Renews Dedication to Advocacy

By Stephanie Carmody

Growing up we called it “Steffi disorder.” My friends and family were as baffled as my expert neurologists. I had been diagnosed with typical Spinal Muscular Atrophy (SMA) as a toddler but never followed its progression; I never seemed to get weaker. My myriad of symptoms was distinctly different than anyone else’s I had ever met in a lifetime living in the neuromuscular community. I thought I might never find my true diagnosis, let alone others who share it with me.

Through the years, the need to find an answer led me to my own online research searching for any disorders that seemed to fit my symptoms. I had severe lower leg weakness and foot deformities from birth, partial vocal cord paresis, scoliosis, hand weakness, and respiratory weakness with severe sleep apnea that developed in adulthood. Along with this weakness, however, I maintained very strong upper body and hip muscles which allowed me to walk with KAFO’s and Iofstrand crutches into my mid-twenties. I had never been able to bear weight on my legs to stand, but when I received my KAFO’s and walker at 18 months, I took off walking and only grew stronger. Up until last spring, no diagnosis seemed to fit.

My life was forever changed when I came across a blurb about newly recognized atypical types of SMA not related to the SMA gene. After researching further, I discovered that a gene named TRPV4 was found to be related to three neuromuscular disorders in 2009, 1) Charcot-Marie-Tooth (CMT2C), 2) Scapulo-peroneal Spinal Muscular Atrophy, and 3) Congenital Distal Spinal Muscular Atrophy. When I found the list of symptoms related to these disorders I was flooded; they matched mine almost perfectly. I learned that these

three disorders were now considered one disorder, TRPV4 Hereditary Motor and Sensory Neuropathy (TRPV4 HN), with a spectrum of severity.

Cautiously optimistic, I asked to be tested. Two weeks later I had the answer I had been seeking for 34 years. The results came back positive. I was overwhelmed with gratitude and excitement; I finally knew what was going on in my body and for the first time had a tangible avenue for both finding others who shared my diagnosis and advocating for treatment.

At the time of my diagnosis, my genetic counselor said there were only 25-30 documented cases in the world, with only one other individual who shares my exact mutation. Faced with such a rare and newly identified disorder, I realized that the challenges for us as a community were going to be finding each other, raising awareness of our existence, and advocating for research into treatments. I couldn’t help but find it my mission.

To begin these efforts, I turned to social media and posted in groups related to CMT and hereditary neuropathy searching for others who had tested positive for TRPV4 HN. When I found a few others, it was thrilling to finally be in contact with people who understood many of my experiences related to our disorder. I was not alone. I started a private Facebook group for us to share experiences, research, and resources specific to our disorder. Our group has now grown to include eight of us who share our diagnosis. I anticipate that the group will grow as more and more people previously misdiagnosed or undiagnosed undergo genetic testing



and test positive. As far as I know, our “gathering” of individuals with TRPV4 HN is the only one in the world.

Since TRPV4 HN is very debilitating and can be life-threatening because of respiratory involvement, the need for research into treatments is critical. It is a disorder of the calcium ion channels, and research is hopeful for treatment because the gene and its function are so well known, and preliminary research indicates that some of the muscle weakness may be reversible. The challenges now are identifying patients by advocating for genetic testing for previously undiagnosed or misdiagnosed patients, and raising awareness for the need for research to investigate potential treatments.

What a difference a year makes; from “Steffi disorder” to a definitive diagnosis with hope for potential treatments. When I look to the future, I am hopeful for answers to the remaining scientific questions. With partners like the Hereditary Neuropathy Foundation and Biopontis Alliance for Rare Diseases, I envision a world where, through treatments, individuals diagnosed with TRPV4 HN won’t have to live with its consequences.

Stephanie lives in North Carolina with her husband and two cats. She has a Master’s Degree from UNC-Chapel Hill, and works in the healthcare field.

Q&A with Timmy Dixon, a Bilateral Amputee with Charcot-Marie-Tooth (CMT1A)

By Courtney Hollett
Fundraising Coordinator – HNF

COURTNEY: Tell us about yourself.

TIMMY: I was born and raised in a small town in the Blue Ridge Mountains where everyone knows your first name. It is very laidback and quiet, no stoplights, no hustle and bustle, and no rush whatsoever. I am a standout in my town because I am unique and have a story to tell.

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

TIMMY: When I was growing up I always had that feeling something was different about me. I tried very hard to fit in with my friends and that was a huge challenge for me. I was very clumsy growing up and found it difficult to even walk a straight line. As a kid you are usually pretty reluctant to visit the doctor, but my parents made me. At the young age of 14 I found out I had CMT. A neurologist administered an electromyography (EMG) to check the speed of my nerve conduction. As suspected, I had CMT. My dad suspected I did because he had CMT as well and struggled daily to make the best out of life. I was very young, scared, and in denial choosing to ignore the disease and try to fit in with everyone else.

COURTNEY: What were the events that lead to bilateral amputee?

TIMMY: As I grew older things just became more difficult to accomplish and life was becoming overwhelming. Daily tasks as simple as walking, tying shoes and the ability to stand for over 10 minutes became almost



impossible. I had to earn a living out of high school so I tried my hand at various places of employment. I would last a few months, if lucky, and would have to return home unable to take the pain of standing on my feet. There was always a hopeless feeling deep inside me which is hard for me to deal with growing up. Due to making some bad decisions and having financial burdens I was no longer able to keep valuable doctor appointments and checkups. In January 2012, I was rushed to the emergency room and in critical condition. I had been battling foot ulcers and infection for the past 5 years and I was in danger for my life. I was sent to Duke University Medical Center where the preformed emergency surgery to remove both of my infection riddled feet. This surgery was just the first of a series of surgeries I would have to endure in the upcoming months. The surgeon came to talk to my family members when the procedure was over. He told them I was in serious need of action, if I had been 1 day later I would have been a dead man. After post-op infections, contracting MRSA (a life-threatening

staph infection), and 4 surgeries later, I am a bilateral below the knee amputee (BBKA). The struggle of phantom pain and wrestling with my prosthetics every day is really trying to me, but I have to keep pressing forward. I have a cure to find and people to inspire, I cannot just quit now!

COURTNEY: What are the challenges you face with CMT?

TIMMY: While I was in transition to my new life as an amputee I was very unhealthy and overweight. The physicians expressed their concern to me before I came home from the hospital. They told me it wasn't going to be easy for me carrying all the extra weight around so I decided to make a change, a chance at a better more active life. I have always heard that a good diet and exercise is the key to weight loss, and here I sat with no legs totally non-weight bearing while I recovered. In 7 months, I managed to lose 125 pounds by changing my eating habits. I felt the healthiest I have ever felt in my life. Losing the weight was also a big benefit to my affects

from CMT helping me accomplish goals that were merely a dream of mine. I hear people tell me all the time how they would love to lose weight but they are in too much pain. Imagine not having any legs and just endured major surgeries and still accomplishing your goals for a better life. My CMT is still very visible and a daily struggle of mine but, I have to keep pushing forward. I am here for you and offer myself to help you reach your true potential, despite the physical altercations you may be facing. Rome wasn't built in a day and neither are you, it takes a lot of time and patience to overcome what lies ahead.

Meet Team CMT Michael Watkins From Braselton, Georgia

By Courtney Hollett
Fundraising Coordinator – HNF

Michael lives in Braselton, GA and is married with two great kids. He is an avid cyclist and decided to go out on a limb and throw his name in the hat for a lottery trail race in Leadville, Colorado. The Trail 100 MTB Race which will take place August 15, 2015. This trail race is widely considered one of the toughest mountain biking races in the country. The race is 103.3 miles long, take place at elevations ranging from 9,300' above sea level at its lowest point to 12,550' at its highest point and makes its riders climb a total elevation gain of 12,612' all in under 12 hours. 100 mile trail races are difficult, but imagine having CMT? We are so grateful that Michael reached out to us to cheer him along the way as he tackles this amazing feat!

Michael's motivation is to show everyone that having CMT does not mean you can't achieve things you and others once thought were impossible. "CMT may limit what my body can do,

COURTNEY: How has CMT changed your life?

TIMMY: CMT has changed my life in so many ways but I always look for the positives and not the negatives. I feel I have grown and became a more caring person. I have humbled myself because I do know the struggle of living with CMT along with amputation. Life is short; you go around this world one time, make it count and live your life to the fullest. Help your brothers and sisters every chance you have because you could very well be there light of hope.

COURTNEY: How were your feelings leading up to the TD Five Boro Bike Tour?

TIMMY: I have to admit I was a little nervous but excited to ride on behalf of HNF to help find a cure for CMT. Finding a cure for CMT has become a passion of mine and I will grasp every opportunity I am presented to help make a difference. I see great things happening for us in the future but now, more than ever, we need to come together and fight together. I have faith in each and every one of you.

It's not too late to **sponsor me** to reach my fundraising goal!



but it will not and cannot limit my goals, my will and my determination to bring a finishers belt buckle back home"!

Michael trains 6 days a week and has many hours to think. He stays motivated during a tough day of training by repeating this motto to

himself "sometimes we just have to work a little harder than most to get what we want and the harder you work at something the greater the feeling you have when it's over".

Follow Michael's blog and consider donating to his goal!

Run Like a Superhero to Benefit Autosomal Dominant Optic Atrophy (ADOA)

By Courtney Hollett
Fundraising Coordinator – HNF

On Saturday April 18, 2015, hundreds of people hit the pavement dressed like their favorite superhero to raise awareness of a rare disease called Autosomal Dominant Optic Atrophy (ADOA). ADOA affects 1 in 50,000 people and mostly strikes in early childhood.

Carolyn Nava organized the Superhero 5K in honor of her 4-year-old daughter Jillian, who was born with this rare disease. Currently, there is no cure – and until one is found, Jillian will soon lose her abilities to see and hear.

The race was followed by a family-fun day in Lexington's Virginia Hilton Park, with local artists, a balloon maker,

vendors, food, and much more. A DJ rocked the house to keep the runners and walkers moving. The highlight of the day was the guest of honor herself, Jillian, as she showed off her dance moves. At the end of the day the event was a success raising over \$9,000. Plans are to make this an annual event.

In addition to this, Carolyn will plan on coordinating multiple events in South Carolina to continue raising awareness as well as funds research. For example, she and another two families that have children affected with ADOA have teamed up by organizing a virtual Superhero 5K. Runners can run anywhere, any day between April 20th – June 30th. Please help these families reach their goal of \$50,000 by December 31, 2015.

Click here for information and to register. Remember that every dollar, no matter how small, can help prevent these kids from losing their eyesight.



Team CMT Takes On the TD Five Boro Bike Tour

By Courtney Hollett
Fundraising Coordinator – HNF

On Sunday May 3rd, 2015, twelve Team CMT members biked through the five boroughs of New York City on streets totally free of cars. The day was picturesque with not a cloud in the sky. One member that had never been to

New York City stated "I definitely felt the love this weekend in New York City." These twelve individuals were raising funds for HNF's Therapeutic Research In Accelerated Discovery program (TRIAD). We are thankful for our riders and donors for their continued support as we raised \$20,000 for CMT research.

Click here, it's not too late to donate to the TD Five Boro Bike Tour!

"HNF is so grateful to the TD Five Boro Bike Tour for their continued support year after year."

– Allison Moore
Founder & CEO – HNF



UPCOMING EVENTS

Save the Date

The Virtual Superhero 5k
April 20 – June 30, 2015
Anyplace you want

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

Dick Sharpe Memorial Golf Outing
Thursday, September 17, 2015
Plandome Country Club
Manhasset, New York

Grace's Courage Crusade
Sunday, August 16, 2015
Narragansett, Rhode Island

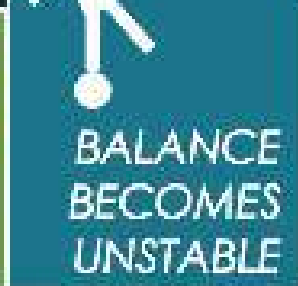
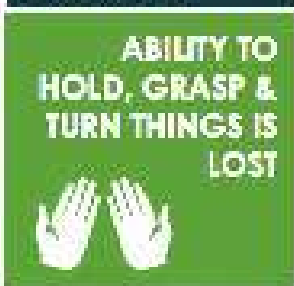
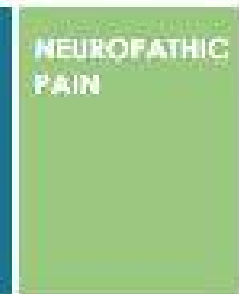
Pizza With a Purpose
September 14 – 16, 2015
New York, New York

Spin NYC at Crank
Fall 2015
New York, New York

For more information or questions about upcoming events, please email Courtney@hnf-cure.org

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

everything seems ok until her...



Even if she is lucky enough to get a **proper diagnosis** of this genetic disease, there are **no real treatments** & it is **progressive**. The outlook can be bleak.



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New York, NY 10016



@CMTNeuropathy



HereditaryNeuropathyFoundation

CHARCOT-MARIE-TOOTH DISEASE

CMT

**IS ONE OF THE MOST COMMON INHERITED
NEUROLOGICAL DISORDERS**

AFFECTING APPROXIMATELY

1 IN 2,500

PEOPLE IN THE UNITED STATES

**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.



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Find out more at hnf-cure.org/cmtprovider