



# CMTupdate™

Spring 2014



Dr. Andrew Grierson

## Using Zebrafish to Search for Therapeutics for CMT2A

SEAN EKINS, Phd, DSc

Research on CMT is global, and covers both laboratory and clinical studies. It is critically important to be aware of what is happening elsewhere as well as in the USA because it can have implications for what we do and fund at the HNF. In June of 2014, a paper was published in PLOS ONE by Chapman et al. (from the laboratory of Dr. Andrew Grierson at the University of Sheffield, UK) that caught our attention.

While we frequently see researchers using mice and rats as animal models of diseases, sometimes they are unsuitable for knocking out genes due to embryonic lethality. Many researchers have turned to the humble zebrafish to further their understanding of genetic disorders, and this investigation was no exception. By using the zebrafish as a model of CMT2A, (which affects the distal axons of motor and sensory neurons in humans), the study was able to examine how the mitofusin 2 (MFN2) affects the phenotype in the zebrafish.

Using zebrafish in genetic research is an active field, but there have been some limitations on the availability of genetic models of diseases. Genome editing technology, however, has developed rapidly, so it is now quick and cheap to make mutations in almost every gene now. Screens in fish are longstanding for studies of development. The MFN2 fish doesn't get a phenotype until it is older, so it can be a much harder task.

Almost all human disease genes have an equivalent in zebrafish. Zebrafish are Vertebrates (as they have a spinal cord), which is critical for modeling disorders of the motor system such as CMT. Zebrafish are highly fertile and it is possible to generate hundreds of fish bearing the CMT2A mutation in a short time. Testing can

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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.

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then be done to measure the effect of various therapies and see which ones show the most promise for future use in human CMT patients.

As the mitochondria are the energy source of the cell, any gene mutations that affect their function can have far reaching implications for health and can cause CMT2A as well as other neurodegenerative diseases like Parkinson's disease. The zebrafish are frequently used as a model of human diseases of the nervous system such as ALS and spinal muscular atrophy. While zebrafish created with this mutation in MFN2 initially developed normally, they showed progressive motor dysfunction between 100 and 200 days old. Some patients with mutation of the MFN2 gene also show progressive motor dysfunction. In the study, fish were monitored by video in their aquarium and those that swam at an angle of more than 30 degrees to the horizontal were recorded. In vitro cell culture was used to measure mitochondria transport in the neurons from the MFN2 knock out zebrafish and retrograde transport was decreased.

While humans and zebrafish certainly do not look alike, their mitofusin 2 proteins are very similar. This study presented a very useful animal model of CMT2A that can be explored further to test potential drugs.

We hope to report on future developments from this laboratory as the HNF is contracted to support targeted therapeutic experiments for one year.



## GLOSSARY OF TERMS RELATED TO CMT TYPE 2 AND ZEBRAFISH RESEARCH

**Mitofusin 2 (MFN2):** The protein that in humans with CMT2 is encoded by the MFN2 gene.

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

**Phenotype:** A description of your actual physical characteristics. For example, the symptoms of CMT 2A manifested in the zebrafish or in a patient.

**Axonal Transport:** A cellular process responsible for movement of mitochondria.

**Mitochondria transport:** Refers to movement of the mitochondria in the axon.

**Retrograde transport:** Backward movement of mitochondria.





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

## Help us answer questions that your doctors and the CMT Research Community aren't too sure about.

Did you know that you can become part of a community in therapy development and further research for all forms of CMT and inherited neuropathies? The mission of the Global Registry for Inherited Neuropathies (GRIN) is to collect clinical and genetic information from patients with ALL forms of Charcot-Marie-Tooth (CMT) and other related rare and ultra rare inherited neuropathies.

We are actively seeking patients with a wide variety of symptoms: numbness or tingling, foot drop, hand deformities, muscle atrophy and weakness, as well as more unusual symptoms of hearing loss, breathing problems, and impaired vision. By contributing your information to GRIN, we can identify trends or find areas of these diseases that may not be recognized. A primary goal is to analyze the data to find patterns and the effects of living with a progressive neuromuscular disease. This will give us more insight of how your particular mutation affects you and others. More data will help us quantify your answers to some of the most important questions in the drug discovery

process. The experts and stakeholders require this information to make breakthroughs in therapeutics and drug discoveries.

**Please enroll today...it will only take 30 minutes or less to complete your profile. By contributing your information to GRIN, you will:**

- Progress research to advance understanding of and treatments for inherited neuropathies
- Make a difference in the lives of others with inherited neuropathies
- Be informed of the latest clinical studies and trials for inherited neuropathies

Your profile will remain anonymous, so any information that you submit will not be tied to you directly, just added to the registry of information for analysis. Make a difference in the future treatments and understanding of inherited neuropathies...please join GRIN today!

## RESEARCH PROGRESS - THERAPEUTIC RESEARCH IN ACCELERATED DISCOVERY



### DRUG DISCOVERY



### DRUG DEVELOPMENT

Discovery Biology	Translational Medicine	Clinical Trials	Patient Outcomes
<ul style="list-style-type: none"> <li>▪ CMT 1A assay development</li> <li>▪ CMT1A/1E high content screens</li> <li>▪ CMT 2A mouse model</li> <li>▪ CMT 2A assay development</li> <li>▪ CMT2A low content screens in zebra fish</li> </ul>	<ul style="list-style-type: none"> <li>▪ "Proof of Concept" studies on an in vivo mouse model with CMT 2A</li> <li>▪ Transgenic CMT 1A rats for testing various therapeutics</li> <li>▪ Partnering with pharma/biotech (in process)</li> </ul>	<ul style="list-style-type: none"> <li>▪ Partnering with pharma/biotech (in process)</li> <li>▪ New outcome measure/biomarker for CMT1A/CMT2A (grant submitted)</li> <li>▪ Alternate outcome measures for CMT (grant submitted)</li> </ul>	<ul style="list-style-type: none"> <li>▪ Global Registry for Inherited Neuropathies</li> <li>▪ Diagnosis Patient education (Quest/ Athena Diagnostics)</li> <li>▪ Quality of Life study (grant submitted)</li> </ul>



## A Birthday Wish!

On Saturday March 1, 2014, the Espace club in New York City was the setting for an amazing party to celebrate Hereditary Neuropathy Foundation (HNF) supporter and past board member Marguerite Loucas' 50th birthday. Guests indulged in the exquisite spread of appetizers to desserts and danced the night away.

Marguerite participated in HNF's newest endeavor, "Birthday for a Cause". She gathered hundreds of her friends and family and encouraged them to give to a cause that she holds dear to her heart: HNF. Guests were asked not to shower her with gifts, but to give to HNF in honor of her birthday wish. HNF is grateful for Marguerite's generosity, as her guests have donated over \$7,000 to fund CMT research to date. Thank you to Marguerite and her family and friends for making a difference in the lives of ALL of us affected by CMT!

Please consider having your own "Birthday Wish" to support CMT Research!  
For details go to [hnf.donorpages.com/Birthday/](http://hnf.donorpages.com/Birthday/) or email [Courtney@hnf-cure.org](mailto:Courtney@hnf-cure.org).

## Milestones & Memories

Planning a wedding is a time of endless decisions. Days are filled with thoughts of invitations, flowers, photographers, wedding venue and countless other details. Hereditary Neuropathy Foundation supporters and Team CMT members Cheryl Monnat and Robert Kearney made a special decision when planning their May 24th wedding. They decided to ask for donations to HNF instead of gifts. It is a wonderful gesture to turn their wedding into a chance to help those with CMT. They have set up a fundraising page on the HNF site for friends and family to mark their special occasion.

Thank you Robert and Cheryl for your thoughtfulness and generosity. Most of us have enough stuff in our lives. Wouldn't it be wonderful to mark a special occasion like a birthday, retirement, or anniversary with your own fundraising page?

It is easy to do. Just contact [Courtney@hnf-cure.org](mailto:Courtney@hnf-cure.org) or call 919-824-7260 to set up your page. Once your page is active, email your friends and family to tell them about your own memorable milestone.

Make it one with lasting impact for those affected by CMT.



# Team CMT & AWARENESS

## From the words of a CMT Hero

### “FAITH, LOVE AND HOPE KEEPS TIMMY DIXON GOING!”

I often wonder what it's like to be like everyone else out there. How great it would be to do things most consider easily accessible and within reach. I believe in myself; it's not really about that. Sometimes I just feel alone, cold, and bitter after considering life in general. Then I ponder a thought: there are too many hurdles I've already jumped over. I always seem to get up off the ground and dust myself off. I realize I am either dumb or tough or maybe, a little of both. I feel as if I have seen a lot in my 35 years, and the years have broadened my mind both in endurance and faith, both which I find very useful. When I was a young boy growing up in a small family I noticed I never quite fit the average status so many of my friends fell into. I found things would take me twice as long to do, and being graceful wasn't me. My father and I shared something that set us apart from almost everyone in our community. We were both living with Charcot-Marie-Tooth disease. Being a young boy stepping into my teenage years, this was very scary for me. I noticed a change in my abilities year after year, but the prime of my youth kept me from seeking further help.

My father wore special orthopedic shoes referred to as “claw boots.” I remember the look of them sent terror threw my mind daily in fear of becoming disabled and left on the sidelines. My youth slowly slipped away from me, and most of my time was spent sitting down or nursing a



pressure ulcer that had developed on my foot. Years would pass, and I continued to work and lived a semi-normal life. My feet suffered a severe case of foot drop, and my hands were becoming increasingly weak. I would fall daily and drop things I had intended to hold on to. Often someone would notice my struggles with walking. I would choose to dodge the question and change the subject to avoid any pity or a medical suggestion. Mentally and physically, this was all

a very heavy load to bear. As I approached my 30's I had noticed big changes in my condition and was scared to death of what was happening. In the meantime, I had met a young lady and our relationship had blossomed into marriage. I was truly the happiest I had been with my present life at the moment. The only thing amiss was my poor health and the reckless upkeep of my body. I had been keeping all of this a secret from my wife in fear of losing her forever, a thought that was also a big part of my fear. She knew deep down that I was hiding a health issue from her, but out of love had never questioned my wellbeing. Late in 2011, things really took a turn for the worst and I found

myself very sick. I knew in my heart I would have to begin to accept help. An infection was circulating in my body causing me to totally shut down. Time was of the essence as I paid a visit to my local emergency room. I was suddenly asked to make a decision that would change my life forever--amputation. The doctors told me that my feet were so infected from my reoccurring ulcers there was no hope in saving them. I wondered if all the years of

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# Team CMT & AWARENESS

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avoiding doctors, self diagnosing and self treating had brought me to this point. Instantly, I lost it, emotionally crying out and asking, “Why me, God, why me?” But there was no time for me to think about what had to be done, and I had to pull myself together quickly. When I woke up after surgery and I looked down, all I could see were bandages and two machines with wires and hoses draped over everything. I couldn’t see my legs at all but I was sure things were different than they once were. I felt fine, just tired and surprisingly in minimal pain.

The team of surgeons told me that in two more days they would do the final surgery on my feet to amputate them. Here we go again, I thought, with lots of emotions flowing through me. But the second surgery also went well, and I was thankful to hear the words of encouragement the doctors gave me. The amputation on the right side left me with half a foot. On the left side the amputation left just the heel, with a bone fusion of my remaining heel.

I endured months of recovery and being non-weight bearing, close to six to be exact. I appreciated every step I could take after I had the pleasure to stand. My prosthetics were custom-made to support my weight and were very cumbersome, but I had missed walking so much, I pushed that thought aside and had a huge smile on my face. I was still burdened by a small sore along the suture line on my right foot that would not heal. Keeping an open wound a lengthy amount of time raises the risk of problems developing, and sure as

that thought replayed in my mind, I became sick with a very high temp and a throbbing pain in my right foot. I rushed to the doctor right away, but it was too late, I had contracted MRSA, a very dangerous infection. The decision was made to amputate below the knee on my right leg. The surgery went wonderfully and I was up and walking in a few months time. I remember the doctors telling me the more weight you lose, the better your life will be. I began counting



calories and within a year’s time, during recovery, I lost 125 pounds. That year I accomplished so many great things for myself that I never thought I could do. I hiked, went mountain biking and fished regularly with minimal problems. Then, out of the blue my left foot began giving me a lot of problems. I had developed another ulcer caused by pressure from the prosthetic. This was the same spot that never seemed to heal completely. After discussing options with my surgeon and prosthetics team I made the decision to have another below the knee amputation. I awoke

after surgery to the sight of no legs below my knees, a rather humbling feeling.

Today I am walking, with Charcot-Marie-Tooth by my side. I also am a bilateral amputee who is active, never wasting a minute of my precious time. Many things have come and gone in my past three years, and they have taught me a valuable lesson in life: never give up. If we honestly don’t think we can continue, we are not looking deep enough. The will to survive is buried

deep within us and sometimes it takes almost losing a grip on life to regain what you want and more. CMT and my amputations have brought me closer to the things that matter the most in life like my family. They are my rock and my number one support system. Second, I rely on my God, because with Him all things are possible big or small. Last I look to my faith, because I never knew I was that strong until I was tested. Ironically, I thank CMT for opening my eyes to see a new world, appreciate new things, and hold dear every moment I am blessed with.

# Team CMT & UPCOMING EVENTS

## Member Alyson O'Connor Won't Let Her CMT Stop Her!

ALLISON MOORE, CEO of HNF

Alyson O'Connor won't let CMT get her down. She was unofficially diagnosed at the age of 10. Her Dad had the same symptoms, but at the time genetic tests were not available. Her diagnosis was based on an EMG. After having her two children, now age 8 and 6, she decided it was important to determine if she definitely had CMT. As suspected, last year she received a confirmed diagnosis of CMT. Currently her kids are mildly affected and her goal is to spread awareness and raise funds for CMT research in the hope of finding treatments and protecting her kids from the potential progression that can lead to disability. O'Connor knows all too well how bad CMT can get, and her goal is to stay active to hopefully maintain her muscle strength and stave off the progression of her CMT. She realizes she is blessed and "goes blockbuster" in her efforts to stay trim and fit. She teaches spin, boot camp, kickboxing, core and yoga classes. Most days are good, but some days she is just wiped out! She says, "It's hard to push through and teach a high energy class." Keep in

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## Meet Matt Downing HNF Board Member & Bike NY Team Member

For the 3rd year in a row Matt will take to the road and conquer a 150 miles on his bike to raise funds for Team CMT. On May 4, 2014 he will ride with HNF in the New York City Five Boro Bike Tour, and then he will ride to Montauk at the tip of Long Island. Matt shares a diagnosis of CMT with his father, sister and three of his children. He is committed to finding a cure for his children and helping to share the knowledge of those who have found effective treatments for their CMT symptoms. He is also an avid golfer and cyclist and strongly advocates the benefits of an active lifestyle for those with CMT. Matt states, "As a member of the Hereditary Neuropathy Foundation (HNF) Board of Directors, I know your contribution will go a long way towards finding a cure for the millions affected by CMT. HNF is dedicated to supporting those living with CMT by raising awareness and money to fund research into new treatments and a cure. All proceeds from this event will go directly to advance this important mission". Please consider supporting Matt Downing: [hnf.donorpages.com/BikeNewYork2014/MatthewDowning/](http://hnf.donorpages.com/BikeNewYork2014/MatthewDowning/)

## Perception Shift

**GINO PRESTIGIACOMO**

Throughout my life many different people have impacted my views on certain things, whether it be how I speak, how I dress, what I eat or even who I'm friends with. I now consider my viewpoint on life much more mature and sophisticated than it was 10 years ago or even 5 years ago. I think more logically than ever before and I am much wiser now because of my mistakes and because of my success. Recently in my life I met another person who greatly impacted my life. Not only has he greatly changed my viewpoint on life but shockingly he is also younger than I am and we haven't known each other very long. His name is Zach Houliares, he is fifteen years old and he has recently been diagnosed with Charcot-Marie-Tooth, which is a genetic disease that affects his nerves and muscles making walking very difficult. That alone is a struggle, but Zach was also born visually impaired and is legally blind. You would think he is held back in life from doing what most kids in high school do, but truth is he can just about do it all. I'll never forget the first time I met Zach. He stood up, gave me a firm handshake, saying, "Hello, Gino, nice to meet you," with a big smile on his face. I couldn't help but give a huge smile in return because it was contagious and I never would have guessed Zach was visually impaired or had difficulty walking. I was informed hours later by his older sister about his disease and some of the obstacles he had to overcome and I was truly shocked. It amazed me that someone could go through with that every day of their life and can still be so happy and

succeed as much as he has. Zach has close friends, goes to school, plays video games, participates in sports, and helps coach the basketball team at his high school and he succeeds in all of them. Not only is he a joy to be around, but also because of his attitude towards life and his accomplishments, he makes me want to be a better person and appreciate how blessed and fortunate I am. One way I changed after meeting Zach was simply by wanting to be a nicer person to every person I come across throughout my life because you never know if someone is hiding his or her emotions or hiding a disability. You may also never know how sensitive one person may be towards a joke or a simple action. I didn't consider myself a mean person before

meeting Zach, but my ambition to go out of my way more often in order to help someone or being more polite has increased. I feel better about myself and feel that I can brighten up the outlook of others by just being a good person, as Zach has done for me. I've heard a quote before that says, "God gives his toughest battles to his strongest soldiers", and Zach is proof of that statement. He is someone who faces struggles every day that most people in the world don't have to and he handles it with poise and

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mind the majority of her class participants are not affected by a neuromuscular disease. She needs to challenge them. Maintaining balance in yoga class can be extremely difficult. O'Connor works hard and has amazing support from her husband Jim of ten years. Their anniversary is in May, and unlike your average celebration of a night on the town, she and Jim are celebrating by participating together in an Olympic distance triathlon in Oregon.

Jim, who has been a runner most of his adult life, encouraged O'Connor to enter a half marathon four years ago, and she took to it. It has been a long process and very frustrating, she shares with HNF. She is competitive and expects to excel as an athlete and at times is saddened when her feet and legs don't cooperate. Her husband and close friend runners are running 7-minute miles, and she is lucky to keep up with an 11-minute mile. And as with many CMT athletes, it's not always apparent that there is a problem. But for her and many, it's a problem and this level of activity can be very frustrating and difficult. She started feeling very down as, in most cases, she would come in last.

As a member of Team CMT, she was supported and encouraged to enter USA Triathlon (USAT) sanctioned events. All USAT sanctioned races must now offer separate scoring and accommodations for Para triathletes. This allows athletes with physical challenges to compete on a level playing field so they can have a fair shot to excel. CMT affected athletes qualify for this program.

O'Connor is excited to get back into triathlons and see how far she can go. The first of the season is The Oregon Dune Tri on May 10th, and she is planning on three to four more throughout the year. By training hard and with determination, she is convinced she'll excel not only as a runner, but also as a leader in increasing awareness and funds for CMT. To support Alyson and CMT Research: [hnf.donorpages.com/TeamCMT/AlysonOConnor/](http://hnf.donorpages.com/TeamCMT/AlysonOConnor/)

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positivity every day like it's no challenge at all. I have yet to see Zach upset or discouraged and when I think about it I can't help but be inspired by him.

Zach can be shy at times and perhaps a little hesitant but his true inner strength came out recently for people all across the country to hear about. Zach manages his high school basketball team; he lives the sport through the players by helping them and observing from courtside. You would think someone who is legally blind has a tough time making a basket, but Zach has conquered even that feat during a live action game. Recently during the Victor Blue Devils last home game the head coach had Zach suit up for a chance to finally take the court and play, and Zach was ready. Number twelve in white took the court with confidence. With under a minute left on the clock, one of his teammates gave him a pass inside the arc. He caught the ball, stepped, shot, and drained it as the crowd cheered for Zach. Victor went on to win and Zach was rushed by all his teammates as the final buzzer went off to end the game. The story blew up instantly and was spread across YouTube, Twitter, Facebook, Rochester News, and many other sources. That one shot can describe a lot about Zach's character. He doesn't show fear; he takes chances and makes the most out of the opportunities given to him, which is how everyone should approach life.

For the people who didn't know Zach before his story reached the news, hopefully they can appreciate now how valuable life is even more so than ever before. It's quick, unpredictable, and everyone receives an opportunity at some point in their lives but it's about what you do during those moments. I don't think I can do what Zach has done which is why I respect what he's done even more. He's younger than me, smaller than me but yet spiritually stronger than me and that alone can drive a person a long way. I wish everyone can meet Zach and hear his story because I believe it can change their perspective on life like how it changed mine. I promise to myself to be more optimistic, more polite, and more helpful to others. My mindset has been different ever since that first night meeting Zach and I will do my best to do for others what he has for me. He alone has changed my life and my newest goal is to be like Zach; I am now a better person.

## Join Zach Houliares' Team

Zachary is an amazing young man who enjoys family and friends. He enjoys being involved with school and participants in the sports programs. This past year he was the freshman assistant football coach, the basketball team manager and now an assistant coach for freshman baseball team. Zach was recently diagnosed with CMT Type 6, a rare form of CMT. This diagnosis came after a lifetime of countless visits to ophthalmologists, neurologists and orthopedics. Zach is legally blind with a vision of 20/400 and suffers

from Optic Nerve Hypoplasia. We have spent a lifetime traveling from specialist to specialist and not one even considered CMT as a diagnosis. It wasn't until a year ago that Zach's pediatrician suggested this disease was a possibility. I had never heard of CMT and once we researched it and found out what it was, we were devastated. Further doctor visits to a geneticist were suggested and it was recommended that genetic testing be performed. Exon Genome sequencing is a diagnostic test that had recently

become available that helps identify the basis of rare and unusual genetic disorders. Genetic testing was able to provide the answers to a lifetime of questions that were left unanswered and undiagnosed. What's so amazing is that this disease was discovered over 100 years ago and there is still no cure. Please consider joining or donating to Zach's Team a [hnf.donorpages.com/ZachsTeam/](http://hnf.donorpages.com/ZachsTeam/) and help us fund research.

## Jillian's Team

Jillian (known to many as "JuJu") has inherited Optic Atrophy/Optic Neuropathy from her paternal side. This condition, also known as autosomal dominant optic atrophy (ADOA)-plus syndrome, involves vision loss, weakness in the muscles that control eye movement (progressive external ophthalmoplegia), difficulty with balance and coordination (ataxia),

hearing loss, disturbances in the nerves used for muscle movement and sensation (motor and sensory neuropathy), and muscle weakness (myopathy).

In most cases, this mutation replaces the protein building block (amino acid) arginine with the amino acid histidine at position 445 in the OPA1 protein (written as Arg445His or R445H). It is unclear why the

R445H mutation causes other features in addition to vision loss in affected individuals. There is no cure and we are working to fund research.

How can you help? Join Jillian's Team and help us raise money to fund research for Optic Atrophy/Optic Neuropathy. [hnf.donorpages.com/JilliansTeam/](http://hnf.donorpages.com/JilliansTeam/)



If you or someone you love has vision loss from their CMT or related neuropathy, please contact us. We are initiating research that may help.

Email us at [allison@hnf-cure.org](mailto:allison@hnf-cure.org)

## Hot topics that have hit Inspire Community!

### 1. BENEFITS OF PHYSICAL THERAPY

**Post:** My Doctor recently referred me for physical therapy because I could no longer put my right ankle on my left knee. That makes it very difficult to put on socks or put lotion on my feet or even examine my feet which we should do with peripheral neuropathy because we could be developing a sore or calluses and not know it. The physical therapist said my core muscles were weak and I could develop back problems, she seemed to think my hip problem was because of the weak core muscles. I was surprised that the core muscles were so weak because I have been doing water aerobics and walking. She gave me 4 exercises to do on the bed. It takes less than 20 minutes to do them. I started right before Christmas. I was absolutely terrible about doing them daily because of all I had going on at Christmas time. Even though it has been a very short time and I was terrible on the follow through I am amazed at how much easier I can get up from the floor or from sitting on low chairs. I would recommend that everyone work on their core muscles. I will definitely be doing my core muscle exercises in addition to my water aerobics and I need to get started on my walking again. With my Helios AFO I can walk 3 miles, without them I can walk 1/2 block ;but that's another topic

**Reply:** Can you describe the 4 exercises that she gave you to do, since they worked so well? Thank you!

1. Lie on your back with your knees bent, arms at your side, Feet on the mattress or floor where ever you are laying. Pull in stomach, think of trying to touch your spine with your belly

button hold for 5 seconds. Repeat 20 times.

2. Lie in same position. Pull in stomach muscles and do a marching motion with your legs. Alternating legs do each leg twenty times.

3. Bridge - Lie on back like in # 1 lift you hips off the bed, hold for 3 seconds. Repeat 20 times.

4. Same position as #2 but as you march also lift your arms right arm, left leg - left arm right leg. Repeat 20 times for each side.

To see more replies visit the Inspire Community. [www.inspire.com/groups/charcot-marie-tooth-cmt/discussion/benefit-of-physical-therapy/](http://www.inspire.com/groups/charcot-marie-tooth-cmt/discussion/benefit-of-physical-therapy/)

### 2. FOODS THAT AGGRAVATE PERIPHERAL NEUROPATHY

**Post:** Interesting article about foods that aggravate Peripheral Neuropathy <http://strongcustoms.com/foods-aggravate-peripheral-neuropathy/>

**Reply:** Thanks for sharing runnergirl1. I am very sensitive to foods and I believe the underlying reason is my CMT. Many neuromuscular diseases affect the gut. Not only do I experience stomach distention, but the foods act as a stimulate and my sleeps are regularly disruptive. I have read several published papers on the benefits of Curcumin (which makes the spice turmeric yellow) for peripheral neuropathies. The spice given to animals with certain forms of CMT had improved functional tests. I happen to love Indian food so I'm ingesting it often. More studies need to be performed on the benefits of food and natural products for CMT and related inherited neuropathies. If anyone has any ideas of foods and supplements that have helped

with their neuropathy symptoms, please share.

To see more replies visit the Inspire Community. [www.inspire.com/groups/charcot-marie-tooth-cmt/discussion/foods-that-aggravate-peripheral-neuropathy/](http://www.inspire.com/groups/charcot-marie-tooth-cmt/discussion/foods-that-aggravate-peripheral-neuropathy/)

### 3. AFO's

**Post:** I live in New Zealand and am having trouble with AFO's. The ones I have are hard plastic and are very uncomfortable. I also have a lot of trouble finding shoes that they will fit into. I hear that the step smart AFO's are very good. I can purchase them here but without trying them. I believe that they will need to be fitted by an orthotist. My question are they good, is it hard to find shoes that they will fit into, and do they need much fitting or adjusting. They are not funded so don't want to waste my money if they are not good.

**Reply:** If you can afford them the carbon fiber AFO's are really superb. I wore them for 3yrs until I had both my ankles fused. I no longer need braces of any kind. But how successful carbon braces will be for you really depends on your condition. With me, my left foot was getting so bad ( foot was being pulled over ) that really no brace would work well. Tried the Arizona without any success. In the end I had my right ankle fused in 2010. And the left 1 year later. Complete success. I know surgery isn't for everyone. That's something to discuss with a qualified orthopedic. But in my case the carbon fiber AFO's worked really well.

To see more replies visit the Inspire Community. [www.inspire.com/groups/charcot-marie-tooth-cmt/discussion/afos-6/](http://www.inspire.com/groups/charcot-marie-tooth-cmt/discussion/afos-6/)



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### Help HNF by joining iGive.com. Don't Forget to Use iGive When You Shop Online!

By joining iGive and downloading the button to your desktop or smartphone you can help our mission. Every time you shop through iGive a portion of your purchase pre-tax will be donated to HNF. Follow these simple steps and you'll be on your way to earning HNF money!

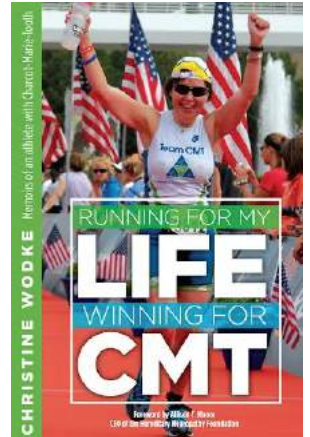


1. Go to: [www.igive.com](http://www.igive.com)
2. Type in Hereditary Neuropathy Foundation
3. Click Join in the upper right hand corner
4. Every time you shop HNF will benefit!

### Support HNF

Running For My Life:  
Book is \$16.95, \$6 of each purchase will go CMT Research. Be sure to use the coupon code at check out: **CURECMT**

[tinyurl.com/CURECMT](http://tinyurl.com/CURECMT)



### Bernadette

Purchase a copy of this film to support HNF and CMT research at [hnf-cure.org](http://hnf-cure.org)

[bit.ly/1rQdjA6](http://bit.ly/1rQdjA6)



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

everything seems ok until her...

 <b>FEET START TO LOOK DIFFERENT</b>	<b>MUSCLES ARE WASTING</b>	<b>SHOES DON'T FIT AS WELL</b>	<b>NEUROPATHIC PAIN</b>
<b>ABILITY TO HOLD, GRASP &amp; TURN THINGS IS LOST</b>	 <b>BALANCE BECOMES UNSTABLE</b>	<b>TOES START TO curl</b>	

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.



# CMT Testing from Athena Diagnostics®

## The Most Efficient Path to an Accurate Diagnosis.



### MOLECULAR TESTING FOR CHARCOT-MARIE-TOOTH DISORDER

Athena Diagnostics now offers advanced sequencing with a cohesive algorithmic approach that simplifies testing procedures and provides the most efficient path to an accurate diagnosis. With a single blood draw, our two-tier methodology first tests for the most common gene mutations based on electrodiagnostic studies and family history. If negative, a second tier test, powered by the advanced sequencing platform, broadens the search to confirm a diagnosis.

To learn more, download our **CMT testing algorithm** at [AthenaDiagnostics.com/cmt](http://AthenaDiagnostics.com/cmt)



Testing that Makes a Difference.

PMP22 | CX32 | MFN2 | MPZ | EGR2 | LITAF | PRX | GDAP1 | RAB7 | GARS | NFL | HSPB1 | LMNA | FIG4 | SH3TC2 | DNM2 | YARS | FGD4 | NDRG1 | TRPV4 | HSPB8 | MTMR2 | SBF2

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