



HEREDITARY
NEUROPATHY
FOUNDATION

What It's Like To Live with Charcot-Marie-Tooth {CMT}

THE STORIES OF THOSE WHO KNOW IT BEST







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A Message from Allison Moore

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One of our most important missions at HNF is to show people what it's like to live with Charcot-Marie-Tooth (CMT). Unfortunately, our disease is still relatively unknown to the general public. As one participant aptly noted, "Because you don't have a broken leg or something readily visible, most people don't understand." Indeed, people frequently ask us questions about our symptoms, our daily challenges and treatment options. And while we try to give them an idea of what it's like to live with a progressive, hereditary disorder, often our descriptions fall short.

To address this issue, we asked ourselves: How can we hope to promote a better understanding of the disease if we can't properly describe it? Our answer was to commission our first Quality of Life research study about what it's like to live with CMT. We were fortunate to draw on the expertise of two highly experienced investigators who conducted interviews with more than 80 people. Our goal was to delve deeper into the world of CMT and to paint a vivid picture of some of the emotional, spiritual and psychological challenges that accompany the physical challenges brought on by this disease.

These interviews brought to light a range of different experiences. They were alternately devastating, courageous, heart-wrenching and inspiring. They told us about the good, the bad and the ugly of living with a disease often not recognized or understood by family, friends and, surprisingly, even physicians. This study illustrates what it's like to live with CMT as told to the researchers by those who know it best.



Living With CMT: The Study

PURPOSE

The aim of this study was to gain a clearer understanding of the experience of living with Charcot-Marie-Tooth (CMT). To date, the research has consisted primarily of quantitative studies. While quantitative research is critical to obtaining knowledge related to more effective treatment outcomes, it does not speak to the experience of what it is like to actually live with this disease. The information within this study, which used a modified Van Kaam approach, goes beyond individual reports, anecdotal evidence and quantitative descriptions to describe the impact of CMT on various aspects of life. Our goal is to help people with CMT learn more about how the disease is affecting others. In addition, we hope to educate health professionals about some of the lesser-known symptoms, emotions and experiences of people with CMT, so they can better understand how to deal with CMT patients and their families.

PROCEDURE

Data was collected using a web-based approach. Participants responded to an invitation to participate that was posted on the website of the Hereditary Neuropathy Foundation (HNF). It allowed participants to complete the questionnaire and submit confidential responses online. The researchers culled salient points from the interviews and combined them here to showcase the highlights.

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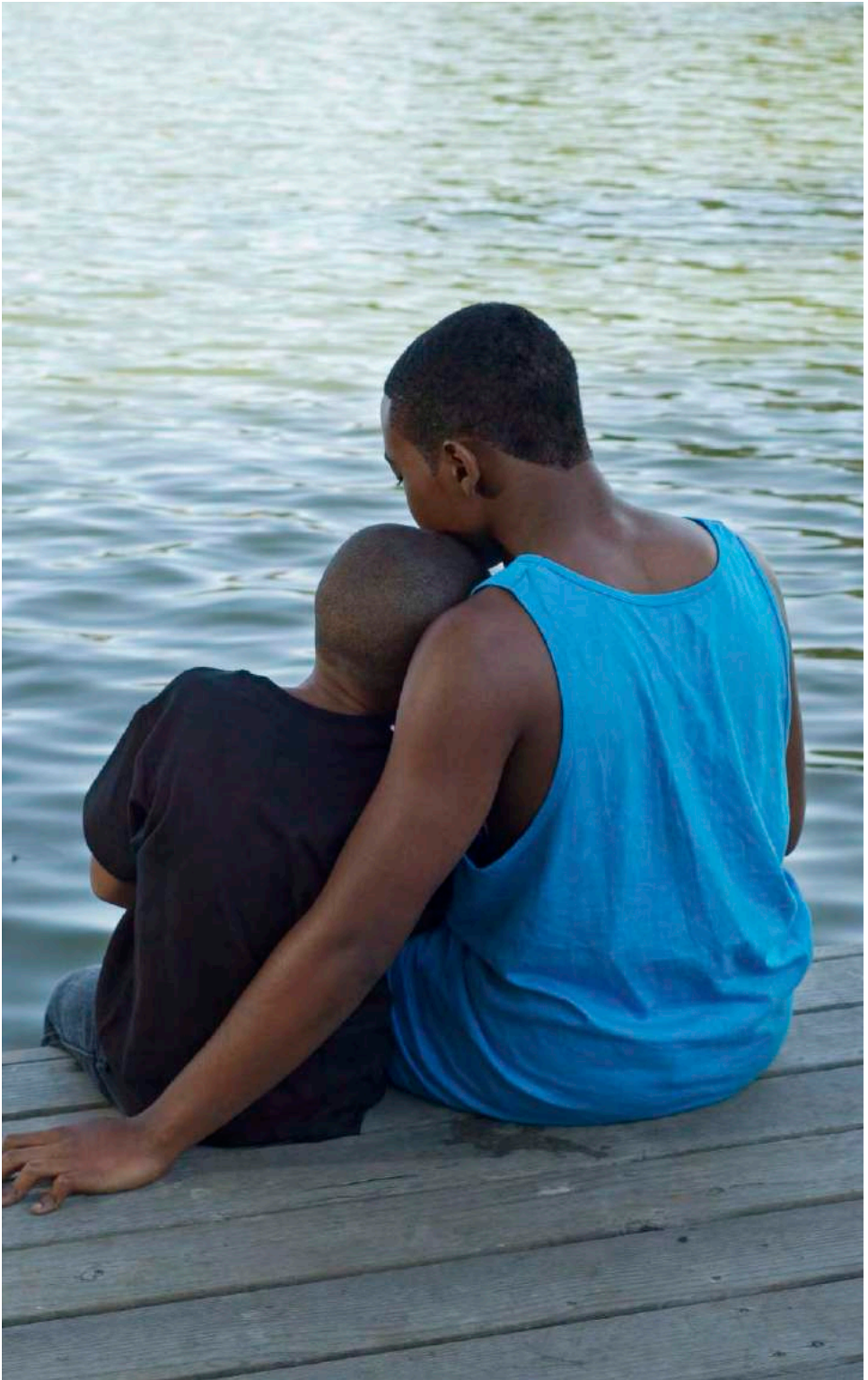
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Research Findings

STUDY QUESTION NO. 1

We asked participants to describe what it is like to live with CMT.

Please share all your thoughts, perceptions and feelings of your experience of living with CMT, in as much detail as possible, and include all aspects, such as the physical, emotional and social effects on your life.

We identified common themes and responses below:

PHYSICAL EFFECTS

People with CMT report great differences in the way the disease manifests itself. Most people reported symptoms progressing

By the middle of the week it was up to my knees, and by the end of the week it was all the way up to my hips. Everything felt numb, but it wasn't. I saw my doctor and told him that my dad had CMT. He didn't diagnosis it, however. I finally found a doctor who did!"

It was clear from the data that age is not a predictor either in onset or rapidity of symptom progression. The following example shows the contrast between mild and severe CMT of two persons, both of whom were diagnosed at a young age. The first one told us, "I was diagnosed as a child. I have grown up with CMT. It is just part of who I am, like me being an artist, a career person, a family member. Physically, I have always been mildly affected. Emotionally, I stay on an even keel. I learned to meditate and that decreases my stress level. Socially, I am a social butterfly." Consider the difference in the response of this person: "I quit school at 16 because I could not keep up with classmates. I wear leg braces and my hands are giving me a hard time. It is hard to get my shoes on and tying the shoelaces is so hard. I drop everything I touch and I really get mad a lot. I am having a hard time sleeping. I can't stay in bed because my legs hurt too much, so I sleep in my lift chair. I don't get out much since my balance is off, and I feel like I'm going to fall."

PHYSICAL SYMPTOMS

Participants reported different types of pain in varying intensity. Other symptoms include:

- » Burning sensation in hands and feet
- » Pins and needles – for minutes or hours

Emotionally, I stay on an even keel. I learned to meditate and that decreases my stress level.

slowly over a number of years. A few, however, had symptoms that progressed quickly as in the following example. "I woke up and my right foot felt like it was asleep, but it wasn't. I had pins and needles all day, and I couldn't figure out why. Then the next day it was in my left foot.



- » Hand cramping
- » Leg cramps/spasms
- » Neck pain
- » Back pain
- » Loss of balance of varying degrees, including wobbling, falling, and tripping
- » Limited ability to walk; sometimes requiring a wheelchair
- » Weak ankles and other weaknesses in extremities
- » Numbness in extremities
- » Clumsiness
- » Decreased ability to stoop, squat, kneel, jump, climb, run, or do anything that requires either upper or lower strength
- » Difficulty grasping and holding objects and opening jars and bottles
- » Increasingly accident-prone (cutting fingers or burning hands while cooking)
- » Itching
- » Sensitivity to cold and Raynaud's Syndrome
- » Restless Leg Syndrome
- » Periodic involuntary limb movement during sleep
- » Foot drop

- » High arches, high instep
- » Hammertoes
- » Small legs
- » Fatigue of varying degrees
- » Progressively worsening gait
- » Progressive loss of function

EMOTIONAL EFFECTS

Depression, with or without treatment by medication, was the most commonly voiced expression of CMT-related emotional pain. Participants rarely mentioned other modalities for treating depression, such as psychotherapy. It is clear that many people with CMT are living with the emotional fallout without emotional or psychological support, which could be beneficial.

Participants revealed many other feelings including disappointment, discouragement, frustration, sadness, feeling defeated, feeling spiritually ill, feeling overwhelmed, loneliness, uncertainty, anger, and fear of being defined by CMT.

For some, it eroded self-confidence. Others noted that family members were "depressed" by or "unaccepting" of the patient's declining physical abilities. A



greater number, however, reported that spouses and others in the family were a strong source of support. Many participants struggled with guilt because others had to take over physical chores, such as housework. Anger often resulted from others implying that the participants were lazy.

One participant said CMT is comparable to having emotional problems, “Because you don’t have a broken leg or something readily visible, most people don’t understand.” Another said, “People don’t understand why I need so many things done for me.” That symptoms are not readily visible was seen as both a blessing and a curse, in relation to how others reacted to them.

One person summed up the thoughts of many when she said, “CMT has no proven treatments, no cure, no definitive prognosis. Every day I think, today is the best physical condition I will ever experience, and often it’s not good enough for my soul.”

SOCIAL EFFECTS

For many, as symptoms progress, social life comes to a near standstill. “It’s difficult to plan social activities as my ability to do things changes with every day,” one participant said. “It’s not something I can anticipate; it doesn’t follow any rhyme or reason. This is the physical roller coaster aspect of CMT.”

The reactions of others impacted many. “People offer advice and it feels like they think I am lazy; like when I have to nap during the day.” One pet peeve: “It drives me crazy when people around me

are in a rush and don’t think about how someone might need a bit more time and space to move.” Another said, “I think one of the biggest problems for CMTers is that most people have never heard of it. If more people knew what it was, more of us might receive compassion and understanding as to why we are limited in what we can do. It’s like living with an invisible disability.”

Loss of friends because of CMT was noted, as well as the value of having caring friends. “I have a small group of friends that are supportive and understanding. They know that there are just some days that I need to be alone, as I am having a difficult time dealing with my situation.” Many participants felt that others simply didn’t understand CMT, especially when CMTers look “normal.”

Over and over, shoes were mentioned as having an impact on socialization, as well as on self-esteem. “I can’t wear feminine, attractive shoes.” “I can’t wear decent shoes, so I don’t dress up at all anymore.” The pendulum swings to more extreme situations effecting independence and the isolation that results from physical limitations due to fear of falling, for example. And finally, at the far end of the continuum, one person told us, “I cannot dress myself, walk, drive, cook, etc. This has severely limited my ‘social’ life.”

Diagnosis

DELAYED DIAGNOSIS BY PHYSICIANS

Participants commonly reported a long delay from onset of symptoms until a correct diagnosis of CMT. Their stories described the emotional difficulties of not knowing what accounted for their physical symptoms. One participant summarized the views of many regarding the implications of delayed diagnosis: “CMT has always held me back from being normal; I just never knew it was CMT. I was slower, weaker and less steady on my feet. In the past 15 years I even quit trying to keep up. It was all I could do to work the jobs I took to support my family and myself. If I had known, I could have made different choices. The fact that I have CMT is

not as troubling in my life at this time as the lack of awareness in the medical community is. I could have been getting treated for over three decades now and maybe not have had to put up with so much pain, confusion and suffering. When I speak of suffering, I mean the pain and sickness I brought upon myself by not taking care of my CMT body correctly. Alcohol, long tiring hours of heavy physical labor, knee and back injuries all could have been avoided by treating my body differently. It makes me wonder how many others are doing this to themselves. How many people could improve their quality of life, their relationships, and their abilities just by getting one doctor who knows something about CMT? These days there are tests that can save people from years of wasted life battling fatigue, pain, feelings of



unworthiness, because they can't figure out why they can't be normal. It is nice to know finally what is causing my physical difficulties. I have quit every job I have held within the last 15 years due to unexplained medical troubles, from knee/leg pains to physical exhaustion with no explanation other than diet from my doctors."

"After eight years I received a diagnosis of CMT. The Multiple Sclerosis scare, the Lupus scare took their toll. I visited a psychiatrist about six years ago because I figured if indeed if I was imagining these symptoms, then I needed to have help so I could resume a normal life. The psychiatrist said it was neurological and my neurologist said it was psychiatric. So, I didn't visit any more physicians for two years. These two years were probably the most peaceful time during my journey."

Receiving validation through a diagnosis was often a relief. This was clearly described by one participant: "It was always evident that something was wrong. I fell down all the time and couldn't do what other children could. As early as four years old, I remember my grandmother lecturing me to put my heel down first and not walk flat-footed. I was considered lazy and lacking motivation because I didn't want to go out and 'play' like other children. I avoided taking gym because I was so clumsy and embarrassed by my skinny legs and ugly ankles. I spent most of my school years in ace bandages for sprained ankles. I couldn't roller skate, ride a bicycle, ice skate, swim, run, sit in a way that allowed me to play jacks, or take part in any team sports. I was actually relieved when, at the age of 32,

I was diagnosed by a podiatrist, who was removing calluses from the balls of my feet, as having a classic case of CMT. For the first time in my life, I felt blameless. All of a sudden, something else was to blame, not me. I never resented CMT, because I know it's not my fault that I have it. What I hated was feeling like I was doing something wrong, like I was to blame for being shiftless and lazy. The intimation was always there that I could have done better. Finally, I knew I couldn't."

Another participant said, "It has been pure hell! I was diagnosed just over one year ago. Until five years ago I was

It is very frustrating trying to find doctors who even know what CMT is, let alone believe you when you tell them how much pain you have.

a professional athlete. Now I can't even hold a ball because 'it' has progressed very quickly. In the beginning, I went to back doctors complaining of foot and leg pain. They performed a laminectomy claiming it would fix all my problems. WRONG! Since then I have undergone 12 surgeries on both feet to help relieve pressure that was creating unhealable foot ulcers. Luckily I found a podiatrist/surgeon and a neurologist that have been my 'angels on earth'."

LACK OF KNOWLEDGE BY PHYSICIANS

Many participants complained about the lack of awareness regarding CMT in the medical community. “It is very frustrating trying to find doctors who even know what CMT is, let alone believe you when you tell them how much pain you have. I finally gave up on them. I don’t understand why doctors cannot be made aware of CMT and ALL of what it can cause! I just recently found out about medicines that can be toxic to people with CMT!”

Another participant said, “I have been going to various doctors and getting no answers.” Yet another commented, “I spent eight years battling doctors telling me that they couldn’t find anything wrong; even now, I feel like I have to deal with this frustrating disease on my own.” Yet another one noted, “Neurologists treat people with CMT as if we are all within a box of treatment. Every one of us is different in how CMT affects us.”

INCORRECT DIAGNOSES

Several participants, even those with long-standing symptoms, reported vague and incorrect diagnoses by doctors, including neurologists. Some participants said that they were initially diagnosed with peripheral neuropathy of unknown origin or impaired peripheral nerve response. The situation was sometimes further complicated when people had additional diseases (multiple diagnoses).

One person was misdiagnosed with Multiple Sclerosis five years before CMT was identified. Another, who also noted it

took five years to get a diagnosis of CMT, was told she was faking some of the symptoms in order to get disability benefits.

Interestingly, many arrived at the possibility of having CMT by themselves. “Even though I have not been diagnosed with CMT, it sure looks to me like this is a strong possibility. I located the HNF site tonight by searching for information about ‘hereditary neuropathy.’”

In addition, medical prognoses were not always correct. For example, consider the following participant’s experience: “I was diagnosed in 1974 at a major medical center, and at that time very little was known about CMT. I was 14 years old and was told I would graduate from high school in a wheelchair and be in a nursing home by my early 20s. This has not happened and, at age 43, I am in better shape than I was at 14. When I was given this diagnosis by a noted neurologist, it devastated my life and the life of my parents and siblings. I cried pretty much continuously for two years. I consider myself very lucky and I log onto a CMT bulletin board almost every day, and everyone there is much worse off than I am. I fend off questions all the time, “What is wrong with your leg?” or “Did you hurt your foot?” I stop this with the response, “I have muscular dystrophy,” which of course I don’t, but most people understand this response and will leave me alone.”

These examples reflect only the perceptions of those who described them. They cannot be considered all-inclusive, but they were more commonly reported than were positive experiences.



Attitudes and Coping Methods

Attitudes ran the gamut from primarily positive to occasionally negative and, interestingly, did not seem to be directly related to the severity of symptoms. Understandably, there seemed to be an overall deterioration of a positive outlook on life as the disease progressed over time. Other themes included taking comfort from spirituality as well as having a sense of humor, including laughing at oneself. For one, “CMT is a lonely disease.” For another, “living with CMT is an adventure.” One participant sums up the complexity of attitudes that reflect the experience of living with CMT. “You learn to be a compassionate person towards others, but at the same time you fight with the anger that lies deep within your soul for not being able to be normal!”

Here are a few examples of participants’ attitudes towards living with CMT:

POSITIVE ATTITUDES

“I am persistent, tenacious, stubborn, relentless, and very determined.”

“Whether or not I end up completely disabled is irrelevant. Life is a path with wonderful and frightening sidetracks and junctions. I mourn everything; I regret nothing. I move forward with the grace of my family and my own strength. I don’t know how I came to this acceptance other than faith in my husband, my wonderful family doctor and myself. My life is blessed.” (This was a person who described severe symptoms).

“CMT has made me determined to take care of myself as best as I can.”

“I don’t let uncontrollable things like CMT change my attitude.”

“I have two choices; 1) I can feel bad for myself or 2) I can live each day to the fullest and enjoy all the experiences and tribulations that I may encounter. I choose two.”

“CMT is just part of my life and the



quality of my life is what I make it.”

“Seize the day, trust God, and help others; that’s my motto.”

“CMT has taught me compassion for others and greater understanding. I have also learned patience. I am a better person for having CMT.”

NEGATIVE ATTITUDES

“Childhood clumsiness, poor handwriting, weakness and fatigue made me a loser.”

“CMT has affected my whole life. At school I was teased, never had many friends, have trouble getting along with others, attitude suffers badly and now I am ashamed to go in public because of poor balance. Also, my feet are horrible looking.”

COPING METHODS

Quite a few participants wanted to educate others about CMT, and to share helpful information, such as the person who wanted to tell “everyone” about the “memory foam pillow” that helped her neck and shoulders, “almost like a miracle.”

This example illustrates the theme of how a simple assistive device can improve quality of life. Others often sought and obtained information about CMT and its management from the Internet.

Of note, a 14-year-old spoke of her self-education regarding CMT through information accessed through the Internet. Many praised the HNF website and other websites related to CMT. Many read research studies and wanted to participate in clinical trials and other types of research. Some were hoping for better surgical procedures to help improve walking. Participants found the Internet to be a valuable source of information for education and self-help, as well as a source of encouragement and hope.

EFFECT ON ABILITY TO WORK AND FINANCES

A number of people talked about having to change the type of work they did, decrease from full-time to part-time employment or take early retirement due to physical limitations. For example, a university

professor had to retire due to CMT limitations but is able to work via distance education and conducts research with co-workers using the Internet.

Others described more serious situations, which ranged from, "I feel like I have doomed our finances" to "I've become a bitter person and have been diagnosed with depression; it's all because I can't work to make a living." Fear of bankruptcy loomed large for a few. One person reported that he had no medical insurance and said, "I simply didn't have money to buy it; I lost my job due to CMT limitations. I am dealing with all the pain without any meds, due to my financial situation."

FAMILY HISTORY AND CONCERNS REGARDING CHILDREN

Many participants were aware of the hereditary aspects of CMT as evidenced by discussion of other family members with

the disease whose symptoms ranged from minor to severe. However, no patterns emerged. One said, "My family is loaded with people afflicted with this disease." Another reported that his family of origin had six children and three of them have CMT. Others noted that no other relatives had the disease; at least not to their knowledge. A mother commented, "For me, living with CMT is not all that bad. However, my four-year-old daughter has it much more severely than I do. And that is really tough, for both of us." A 41-year-old mother, after having symptoms most of her life, was only tested and diagnosed after her 11-year-old son was diagnosed with CMT. She developed depression after the dual diagnoses, but "tries to be strong for him."

Although there are genetic predispositions to various types of CMT, some families may not be aware of this; even when they are, it doesn't guarantee understanding and support for those relatives who have CMT. One person said, "One brother accused me of being drunk all the time!"

A major concern for parents was knowing that their children might be stricken with CMT at some point. One mother was told there was a 50% chance that her children of both sexes would get the disease due to the specific type of CMT that was in her genetic line. Other parents commented that their parenting skills were impacted by their declining physical abilities. For example, one person wrote, "I feel guilty that I can't do physical activities with my children. I want to run and play ball and other games with them, but I can't."



Interventions and Treatments

Reports of interventions and treatments were quite limited as the research questions asked only that participants describe what it is like for them to live with CMT. In a later question, we asked specifically about the positive things that



help respondents to cope and, to this question, they did mention some useful treatments and interventions.

Exercise was the most frequently mentioned activity used to help their physical condition. However, many people were not sure if they were exercising too much or too little or if they were doing the appropriate type of exercise. Some reported resistance training and cardiovascular types of exercise. And a few lamented that they had harmed themselves with exercise before they knew they had CMT. One participant said that her neurologist told her that exercise was a waste of her time.

Useful physical devices included special shoes, as well as high-top running shoes, low boots, shoe inserts, orthotics, braces and ankle foot orthoses (AFOs), grabbers, wheelchairs and scooters. Surgery (feet, ankles, knees, back) and medications such as Neurontin, Clonazepam, Celebrex, Vioxx, and Tylenol were mentioned. Treatment by professionals other than physicians included chiropractors and physical therapists. Very few mentioned use of alternative therapies, such as meditation or yoga.

Some commented on lack of treatments: “If only my town had more resources.” “Having no one in person to talk with or any services in my area to aid in improving mobility or emotional strength makes it all that much harder to get out of bed to face the day one more time, to deal with the pain one more time, to have my dreams dispelled one more time.”



Making Positive Changes

STUDY QUESTION NO. 2

Some people think it is possible to make changes in their lives. Making change involves the ability to make aware choices with the intention of freely involving themselves in creating desired changes. We would now like you to answer four questions about awareness, choices, freedom, and involvement in creating changes in your life.

In relation to CMT, what do you find it is helpful to be **aware** of?

In relation to CMT, what **choices** do you make that make a difference?

In relation to CMT, do you feel **free** to take care of your CMT in the way you want to?

In relation to CMT, how do you **involve** yourself in bringing about changes that you desire?

AWARENESS

In terms of awareness, the most frequent response concerned physical limitations

related to CMT. These included such things as: limitations in walking, others' perceptions of limitations, ways to cope with limitations, acknowledging limitations, physical characteristics of the environments (such as knowing the terrain, so as to plan for mobility), steering clear of fast-walking crowds, available facilities for the handicapped (such as wheelchair accessibility), limitations regarding exercise, avoiding lifting or carrying heavy objects, assistive devices to aid in limitations, tips for getting around and doing activities of daily living, awareness of the body's physical limitations and where the body was located in space (such as where feet are), being on guard to prevent losing balance, progression of the disease, ways to use strengths to balance limitations and avoiding pain by knowing more about what things are harmful or helpful (such as using the weather forecast to avoid treacherous conditions in planning events).

Participants reported avoiding fatigue by resting when necessary, avoiding injuries caused by overdoing, pacing oneself, early recognition of symptoms and their treatment, strength required for various



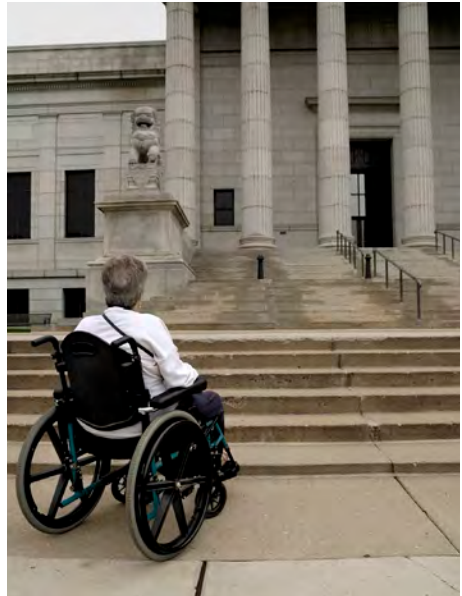
activities, ways to slow progression, and recognition of potential breathing problems.

An awareness of how “CMT can and does change everything in one’s life” was voiced in different ways and often reflected how important a positive attitude was in keeping a realistic perspective. Many talked about the importance of avoiding slipping into the “black-hole of depression” by early recognition of symptoms and efforts to create a meaningful life. Awareness of other’s worse misfortunes in life, and that one bears no “blame” for having CMT helped in efforts to look at the glass as half full, not half empty. Use of prayer and mediation, as well as supportive friendships helped to avoid self-pity. Recognizing that severe episodes will pass helped with endurance. Some reported setting flexible goals to accommodate the changes that CMT brings. Others revealed fears about the changes they will face as they grow older as well as fears of an unknown future.

Awareness of what to avoid was frequently mentioned; for example, drug interactions and chemicals that kill nerves, such as alcohol and other toxic substances. People were eager to learn about new products, strategies and therapies.

Others wanted to learn about new developments in clinical trials, testing, and other research news, as well as CMT in general. Participants’ desire to increase awareness of CMT in the medical world and to gain greater accessibility to experts in treating CMT was loud and clear.

Some said it was their personal mission to get help when needed and help other people with CMT recognize their



rights, enforce those rights, speak up to correct wrongs, help people access care and learn about available assistive devices. One person noted that her father, who has had CMT for 40 years, only recently learned about new braces to help him walk, as well as other programs to help him cope. Another wanted to overcome the fear of asking doctors questions.

In summary, recognizing and dealing with capabilities and limitations, especially as related to physical limitations, was a continuous theme. Searching for ways to educate oneself about CMT, to access quality care, to feel happy and fulfilled despite the disease, and ways to improve quality of life were some of the thoughts, feelings and actions that occupied participants’ awareness.

CHOICES

CMTers in this sample made many choices, often overlapping with their awareness. Many said they choose not to let CMT run their lives, although they were aware of its presence every day. They choose to stay informed by using the Internet and noted that it greatly expanded their choices.

Many actively choose to remain “up-beat” when friends or family inquired about their condition. They made the decision to live fully, not to hide or give up. One response follows. “Choosing to take care of myself has made a huge difference. Saying ‘no’ to certain activities has been hard, but usually worth it.” Another said, “Be happy and if you are not, stop doing what it is you are doing and make yourself a new

as eating habits, conservation of energy, proper fitness programs, family activities and interactions, doing what was necessary even if not desirable, pacing social life and work, and continuously choosing other protective and other beneficial actions in relation to physical limitations.

Others were interested in becoming involved in community activities, taking advantage of available resources, taking charge of health decisions, giving up destructive habits, finding hobbies, making a commitment to altruistic actions for others, bringing information to physicians, avoiding stressful situations, making better choices regarding spending money and making necessary career changes.

While the overwhelming majority of comments were positive, some were negative. Representing the most extreme was, “CMT gives you no choices.” Another said, “I know of no choices I have in relation to CMT.” Some worried that they were not aware of all of the choices available to them.

Reflecting the positive attitude of many, one person said she chose not to be a victim of her genetics. Another told us, “I am a positive, optimistic person. Everything I do is a choice. I choose not to whine and not to listen to whiners. I choose to be a winner and to hang out with winners. I am not a victim of my circumstances.”

FREEDOM TO ACT INTENTIONALLY

The freedom category elicited the fewest responses. In addition, many more people responded “no” rather than “yes.” The

Having sense of humor, choosing to be around positive people, and not dwelling on the disease were some of the more positive coping mechanisms that were reported.

goal.” Many people chose to be around positive people, maintain a sense of humor and, most importantly, avoid dwelling on the disease. It seemed apparent that quite a few were actively choosing positive states.

Many choices reflect the ways they were dealing with physical limitations, such



range was from absolutely no freedom to feeling absolutely free in relation to CMT. These findings are consistent with other research studies on Barrett's theory of power which essentially describes two types of power: power as freedom and power as control. Many people in this study are aware of the choices they need to make, yet taking the necessary action doesn't happen all that easily, if at all. One wonders if this relative lack of freedom is related to participants' general attitude toward making changes in life and whether their attitude shifts as they experience more advanced symptoms.

Feeling free in relation to CMT was most often related to available and adequate medical care and whether their physicians listened to them or were otherwise helpful. Other enhancements of freedom to act intentionally included the ability to overcome embarrassment over using assistive devices,

adequate health insurance, continuously choosing protective and beneficial actions in relation to physical limitations, modalities such as physical therapy and yoga, and financial ability to use available resources to provide care needed for self and family. These resources often were not available, and thus people didn't feel free. Some felt freer as they learned more about CMT and how to deal with it; this gave them hope for management that could slow progression. Some increased freedom through marriage counseling and cleaning up other relationships. Feelings of being stuck in confusion, fear of being open about their CMT after being brought up to hide it, and needing to rely on others for help interfered with feeling free. Some just didn't know what to do and remarked, "The body and spirit are willing, but the mind is in the dark." One person said she regained her freedom by taking charge "since no



one else seemed to have the answers as to how to deal with the CMT.”

One person said, “I feel CMT has power over me, since there is no cure.” The use of a wheelchair helped one person feel free but for another, symbolized a lack of freedom.

There were similar responses regarding use of assistive devices, although most people felt they enhanced their freedom.

Freedom was frequently equated with physical mobility and ability to carry out activities of daily living. One person said, “Of course, I don’t feel free,” but then recounted how she freely acts proactively on her behalf.

INVOLVEMENT IN CREATING CHANGE

Feeling free is the bridge between making a choice and involvement in making the choices happen. What appears as redundancy is actually the difference between the thought (choice) and the action (involvement). We asked participants

to tell us how they involved themselves in creating desired changes related to CMT.

Most participants were very involved in dealing with the changes that CMT has brought into their lives. They struggle daily to function as optimally as possible by limiting activities that lead to injury or fatigue, saying no to self or others when necessary, using assistive devices, asking for help from family and friends when necessary. However, they also strive to take initiative to help themselves rather than relying on others when it is possible to do so.

Some take action by joining groups, such as organizations to help with weight control, to gain support and information, to advocate for better services, or to increase awareness. Reading medical papers, abstracts, speaking in the community, donating to research, and correcting medical misinformation when necessary were ways some of these participants were creating change.

Involvement in finding the best affordable medical care available was an

important activity, as often people linked this directly to making major changes. Other avenues of creating change included implementing treatment plans suggested by physicians, striving for open communication with physicians, and educating them by suggesting treatments. People who had “antipathy toward doctors” because of contradictory or inaccurate advice from them were the exception rather than the rule. Most reported listening carefully to their physicians and then making the final decision regarding their treatment.

Many were thinking about making needed changes but had not yet done so, particularly in relation to exercise and losing weight. Most people tried to deal with issues one day at a time, as they came up. Some struggled with determining what they could change, and what they needed to accept.

Some participants acted on hard choices regarding career changes or early retirement or applying for disability. One person said, “I have formed a support group. I believe that ‘if you’re not part of the solution, you’re part of the problem.’ I try to believe that the things about us that make us the most unique can be our strength rather than our downfall, despite my 18 years of being taught to hide CMT. When I was first told to consider disability, I felt so alone. I couldn’t find anyone to guide me or even to use as a dependable sounding board for my own feelings and decisions. I worked for two more years and I think my CMT got much worse in that time while I was afraid to make the ‘wrong’ decision and ‘let CMT get to me.’

I hope to help others avoid feeling this way; it was awful, and it debilitated me before I even realized it.”

So, here you have it! These are the ways people with CMT feel powerful or powerless. And as previously mentioned, we cannot say anything about people who did not participate. We recognize that this is likely a proactive group, since they took the time to participate in this study. Sadly, few people mentioned the use of alternative/complementary therapies. Those who did mention using massage therapy, physical therapy, yoga, meditation, and chiropractic found them useful. It is recommended that programs be designed to make these services available and to lobby for insurance companies to include coverage for them.





The Last Word: Helpful Comments From Respondents

Three additional exploratory questions were asked.

WHAT POSITIVE THINGS HELP YOU TO COPE WITH CMT?

Helpful positive things identified by participants include use of muscle aides such as massage, muscle strengthening through exercise, stretching, weight training, cold compresses, swimming, yoga and Pilates. Devices such as orthotics, special shoes, canes, AFOs, braces, crutches, wheelchairs and scooters were also helpful. Energy conservation with adequate sleep, avoiding stress, taking breaks, advance planning and schedules were useful. Hobbies, pets, or alternative activities were also described. Other helpful resources: a positive attitude, loved ones and soul mates to enjoy and share laughter and life.

And finally, self-knowledge with active participation on websites and educational forums were viewed as positive coping mechanisms.

WHAT NEGATIVE THINGS HELP YOU TO COPE WITH CMT?

Self-identified negative coping mechanisms used by participants include comfort foods such as coffee, cookies and chocolate as well as use of cigarettes, marijuana or destructive self-medication patterns. Others mentioned failure to exercise and letting stress interfere in their lives. Fears that were viewed as negative include being labeled (a hypochondriac, milking the disease, using the disability for personal advantage, a fat lady in a wheelchair) as well as fear of complaining too much, falling down, and death. Some also experienced guilt about controlling their environment through the use of a “do not disturb” sign or by not answering the phone. One person wondered “if my CMT might be milder if had not consumed alcohol.” People reported

feeling guilty about being stubborn, comparing themselves to others, or denying the disease. Some also reported feeling guilty that others were worse than they were. Finally, anger over lack of control in their lives, leg cramps, years of pain and the medical community's lack of awareness and knowledge of CMT were mentioned.

WHAT ARE YOU NOT GETTING THAT YOU THINK YOU NEED TO DEAL WITH CMT BETTER?

The participants with CMT who participated in this study are not getting the social support needed. They expressed a feeling of social isolation and that no one cares. They need access to other "sufferers," quality support groups and workshops.

People are not getting the help they need to improve their quality of life. A sense of uncertainty and lack of control pervades as they feel there are no options open to them and they worry about what they will face in the future. They cannot plan for the disease. They want to do whatever they can to help themselves but do not know what that is. Worse, they do not know where to go to get information. As a group, they are not getting the help they need to deal with all aspects of insurance coverage especially items covered, Medicare, Medicaid, disability and financial help or support.

There is an appalling lack of information about getting evaluated, where and how they can obtain orthotics, better shoes or help aides/equipment, types of therapies (acupuncture, massage, physical therapy), and help with transportation. Participants

wanted better access to information on pain relief, surgeries, fatigue and helpful medications with low side effects.

Most upsetting to these participants was the failure of the medical community as a source of professional answers. The lack of qualified, interested, accessible care providers is of paramount concern. Access to local care providers and specialists is not available to them. They want someone who is interested in listening to them and who will help them find the best solution for their individual problems.

Most upsetting to these participants was the failure of the medical community as a source of professional answers. The lack of qualified, interested, accessible care providers is of paramount concern.

Study Summary

Keep in mind that this was an online study, so we cannot say anything about people with CMT who do not use the Internet or those who did not participate in this study. However, we can say with a reasonable degree of certainty that for those people who would be willing to participate in a study such as this, the findings would be similar to what we have found. Of course, this will vary by country, culture, type of health care system, and other variables that would be operating in a different research population.

Without a doubt, CMT affects the physical, emotional and social life. The extent of the impact varies with the severity of the illness as well as the attitudes, coping methods, resources, interventions and treatments available to those who knowingly or unknowingly struggle with this disease.

Lack of awareness regarding CMT in the medical community was voiced over and over. These responses illustrate why organizations like HNF are so important in getting the word out as to the real-life experiences endured by people with CMT. Findings of research can be used to increase awareness among professionals, the public, as well as persons with CMT and their families.

It is appropriate to point out that these are stories told from the viewpoint of the people themselves, and their perceptions may or may not be accurate. In no way is this intended to denigrate the importance of medical care; indeed, it points to the need for excellent medical care. Geographi-

cal lack of access to qualified physicians, uninformed physicians, and inadequate medical care for CMT does seem problematic. Although one person told us that a physician had told him there was nothing that can be done to help CMT, others described the benefits they had received from knowledgeable, caring members of the medical profession.

In Conclusion

This undertaking was designed to provide an accurate and immediate picture of what it is like to live with Charcot-Marie-Tooth (CMT). As you have learned, CMT still lurks in the shadows for much of the general public. Unfortunately, a large percentage of those who lack critical information and understanding of this disease are the nation's most revered health care professionals. To the astute physicians, nurses and other practitioners who have read this study, we hope we have put a more human face on CMT so that you will recognize the disease when it is presented to you. We trust that you will be on the lookout for a debilitating hereditary condition that does not discriminate in terms of age, gender, race or ethnicity.

To increase compassion and understanding, we hope that the preceding slices of life have afforded you a glimpse into the often painful, embarrassing and debilitating nature of the disease's symptoms. We anticipate that you will walk away with an increased empathy of daily challenges that CMT patients face and, most importantly, the obstacles CMT patients encounter in



terms of seeking and finding appropriate, considerate and timely health care.

Much of the devastation that accompanies a progressive, hereditary disorder such as CMT is that it is like a time bomb that ticks night and day. For those living with CMT or with the specter that CMT will affect their offspring, the sound can be deafening. Imagine how difficult it must be for these same people to realize that their plight has fallen on deaf ears in the medical community. Our goal is to break the silence.

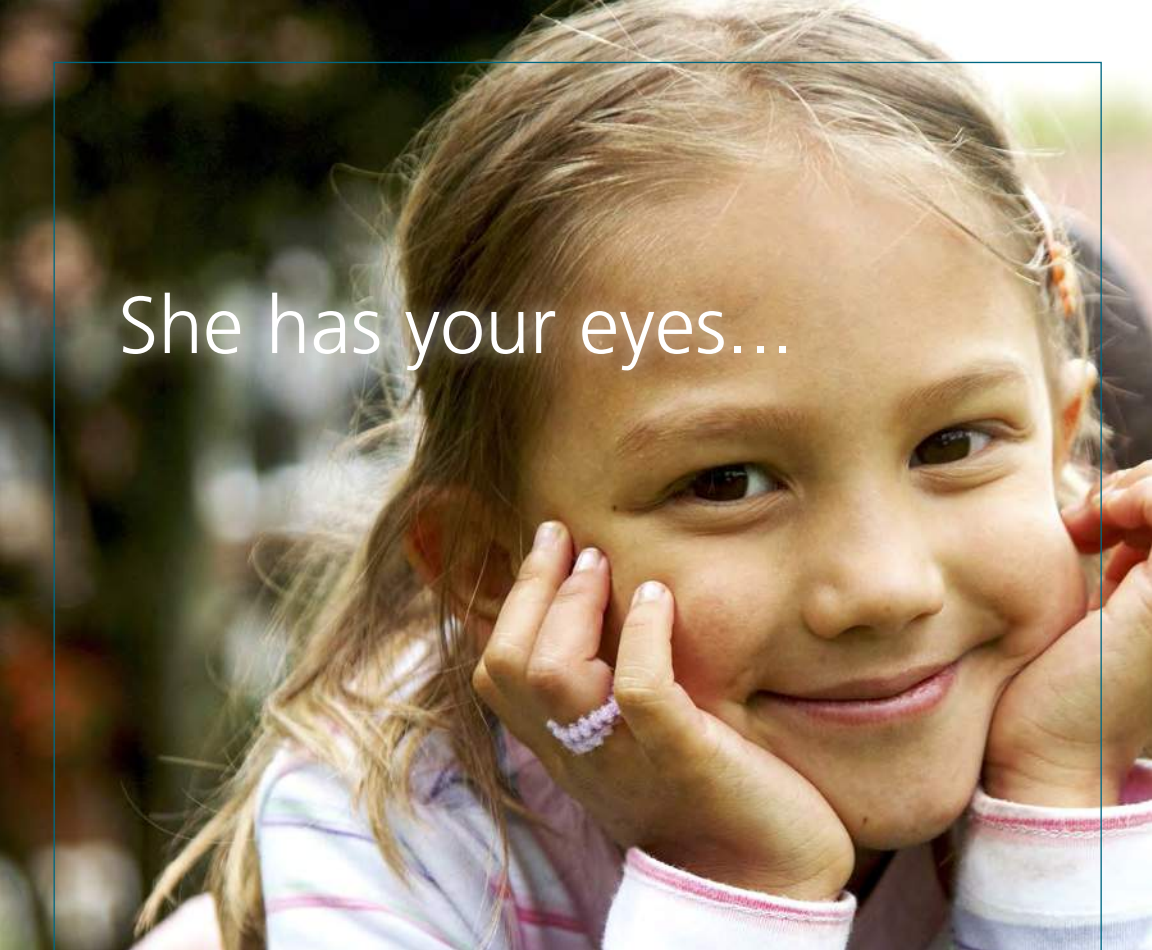
We believe the study has illuminated many of the emotional, spiritual and psychological challenges that accompany the physical challenges brought on by this disease. Your recognition and attention of these challenges will shine the health care spotlight on CMT and bring about a world

of change for people living in the shadows of a little-known disease.

OUR SINCERE THANKS!

The researchers would like to thank the participants for their cooperation in participating in this study. We really appreciate their time and effort and we sincerely anticipate that the findings will be useful to all of those with CMT.

This study was made possible through a grant from the Hereditary Neuropathy Foundation.



She has your eyes...

and your difficulty tying shoes.

It's time to talk about Charcot-Marie-Tooth.

Charcot-Marie-Tooth (CMT) is a progressive, hereditary neuropathy that makes everyday tasks a challenge and can lead to significant muscle loss in hands and feet. The National CMT Resource Center is dedicated to reporting on the latest research and providing essential information and training for families, educators and health care professionals. We believe awareness and open communication are the keys to give those with CMT the invaluable tools for a better life.

One Place. One Vision. One Center.

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This ad made possible by Cooperative Agreement 1U538DD000713-01 between the Centers for Disease Control and Prevention and the Hereditary Neuropathy Foundation.



Therapeutic Research In Accelerated Discovery (TRIAD), a collaborative effort with academia, government and industry, to develop treatments for Charcot-Marie-Tooth (CMT), a debilitating neuromuscular disease. Currently TRIAD involves many groups that span the drug discovery, drug development, and diagnostics continuum.

To learn more, visit <http://www.hnf-cure.org/research/>

To join our patient clinical registry, visit
www.neuropathyreg.org



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