A Guide to Physical and Occupational Therapy for CMT
Charcot-Marie-Tooth disease, the most common hereditary neuromuscular disorder, is named after the three scientists who first reported it. It is also known as hereditary motor and sensory neuropathy (HMSN) because it affects the motor and sensory peripheral nerves. The peripheral nerves are the nerves that connect the brain and spinal cord to the rest of the body. They provide sensory information back to the brain and help control movement and coordination.

People with Charcot-Marie-Tooth (CMT) often benefit from physical therapy and occupational therapy services. This guide can be used by patients and therapists alike to better understand appropriate assessments and interventions that may be beneficial for someone with CMT.

It is important to note that people with CMT are best served by individualized plans of care that are established after a thorough examination performed by a therapist.
Understanding the Cause

CMT is caused by mutations in specific genes that affect the transmission of signals up and down nerves in the body outside of the spinal cord. There are many different forms of CMT; each type corresponding to a specific gene mutation; however, the two most common forms of CMT can be characterized as follows:

Demyelinating forms reduce the myelin sheath, impairing the conduction of signals down a nerve. The myelin sheath is the coating around a nerve. To use an analogy: think of nerves like the wires inside the cord you use to charge your cell phone. The myelin sheath is the exterior, rubbery coating that surrounds the wires. When the myelin covering the nerves is damaged or absent, the messages being conducted are either delayed or cannot reach their destination – in this case, the muscles. Unable to receive the messages, muscles do not function normally and often weaken and atrophy.

Axonal forms damage the nerves themselves. To continue the phone cord analogy, this would be similar to having faulty wires inside the cord that prevent the charge from reaching your phone. Similarly, damage to the nerves prevents messages from reaching the muscles, often resulting in muscular weakness and atrophy as well as loss of sensation.

Signs & Symptoms

CMT affects the longer nerves first; therefore, symptoms tend to start in the feet and hands and work their way up the legs and into the forearms.

Symptoms include:
- Weakness in the feet, ankles and legs
- Difficulty lifting the foot at the ankle during walking (foot drop)
- Depressed tendon reflexes
- Absence of development or loss of muscle bulk in the feet and legs
- An awkward “steppage” gait
- High-arched (pes cavus) feet or flat feet
- Curled toes
- Calluses and blisters on the feet
- Frequent tripping or falling
- Decreased ability to run
- Decreased sensation or a loss of feeling in the feet and legs
- Discomfort/pain in the feet
- Frequent twisting of the ankles and sprains
- Muscle cramping, especially in feet and lower legs

In nerves where the myelin sheath and/or the nerve fiber (axon) is damaged, the nerve conduction velocity (NCV) can be slowed and impulses may not reach the targeted muscles, resulting in muscular weakness/atrophy and sensory loss.
Who Are Physical Therapists?

According to the American Physical Therapy Association (APTA), physical therapists (PTs) are highly-educated, licensed health care professionals who can help patients reduce pain and improve or restore mobility.

In a 2017 survey by the CMTA, 90 percent of nearly 900 survey respondents, all of whom have CMT, reported challenges with mobility. Individuals living with CMT can benefit from having a physical therapist on their health care team throughout their lives.

In the United States, physical therapists now graduate with doctoral-level training. Prior to 2001, many therapists graduated with either a master’s or bachelor’s degree as their entry level degree. Physical therapy degrees are noted as BSPT, MSPT, MPT, DPT or tDPT. In addition to their university training and professional license, physical therapists may also be certified by the American Physical Therapy Board of Specialties as clinical specialists in one of a number of subspecialty areas or hold other degrees (Ph.D., Sc.D., and Ed.D., to name a few). Sub specialty designations include NCS, PCS and GCS. A physical therapist with an NCS is board certified in neurologic physical therapy. PCS means the therapist is board certified in pediatrics. GCS denotes that the therapist is board certified in geriatrics. Physical therapists are licensed by the state where they practice. They may see patients in a variety of environments including inpatient, outpatient, home care, and school systems. These qualifications and degree titles vary with international therapists.

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Who Are Occupational Therapists?

Occupational therapists (OTs) are highly trained health care professionals with a master’s or doctoral degree. They are licensed within the state they practice. They may practice in hospitals, outpatient facilities, schools, skilled nursing facilities, home health care arrangements and private practice. According to the American Occupational Therapy Association (AOTA), OTs help people across the lifespan participate in the things they want and need to do through the therapeutic use of everyday activities (occupations). Occupations represent everyday tasks, including but not limited to:

- work
- play and leisure activities
- self-help skills, including dressing, eating, hygiene and grooming
- fundamental activities of daily living, including cleaning, preparing meals, money management, driving and use of communication devices

What to Expect at Your OT or PT Appointments:

During your first visit, the therapist will conduct an initial evaluation, which will involve taking a thorough history to understand the development and progression of your symptoms. The therapist may inquire about tests or evaluations performed by other health care professionals. He or she will need to know about any other medical conditions or surgeries that may also affect your current situation.

The therapist will conduct a physical examination to document the impact of your nerve involvement on your sensation, muscle strength and flexibility. Physical therapists will assess how these impairments affect your functional abilities with respect to your mobility, balance and endurance. Occupational therapists will focus on how these impairments affect your work and daily life.

The therapist will also assess if functional abilities are affected at all by pain, as well as screen other systems of the body to assess for other problems that may be affecting your functional abilities. Different tools may be used to collect information in a standardized way to monitor your condition over time. This might include strength and sensory testing and measures of functional abilities, such as balance and walking or tasks involving the hands.

When symptoms of CMT affect your ability to work, occupational therapy may be beneficial.
Therapists may also use assessments to determine the impact that your condition has on your quality of life. These assessments provide information on how you are affected by CMT at the time of your visit. The information gathered from your history and physical examination help the therapist formulate a plan of care tailored to your specific needs and goals. Assessments will often be performed again at a later date to determine if progress has been made and to modify the plan if necessary.

After the history and physical examination, the therapist will likely have a further discussion with you and any family members present to better determine your current functional activity level and activities you might feel are limited by your nerve problems. This includes discussing your daily activities and your role in the family and society, as well as your involvement in recreational and occupational activities.

The therapist will then work with you to determine an optimal plan of care. The plan of care is an outline to guide you toward your goals. This plan will include the expected frequency and duration of visits needed to accomplish the established goals. Factors that go into this decision include the complexity of the program, your comfort with your own role and responsibilities related to the program and the therapist’s need to monitor the program.

The therapist will demonstrate and teach you an individualized home exercise program designed to support the activities you do when you come to the office. This will include the expected frequency, intensity and progression of the exercise or activities. The therapist will monitor your response to the program and will make modifications as necessary. Much of the success of any course of therapy rests on diligent follow-through of both the home and clinic-based portion of the established program.

**Goals of Physical Therapy:**
An overarching goal of physical therapy is to maintain or improve your ability to participate in your desired roles. This is accomplished by providing strategies and compensatory techniques to optimize participation. Safety tips and assistive devices, including orthotics, may also be recommended to optimize your function, reduce energy expenditure and maximize participation in daily activities. Your PT may also provide education to you and your family members about the effects of disease progression on function, lifestyle and potential therapeutic interventions. In addition, your therapist may also support realistic expectations regarding functional progression and the impact of intervention over time.

Understanding how strength and flexibility relate to your functional concerns is an important starting point of physical therapy intervention. At present, there is evidence supporting exercise as a way of addressing impairments related to strength. Physical therapists are well-trained in making the best recommendations possible by considering the evidence available, your preferences, and your individual clinical picture. The therapist may recommend a course of care with an intensive clinic-based program followed by periodic visits to monitor your condition, update your home program and make appropriate recommendations for additional therapy services.

**Common Considerations for Patients with CMT:**
While it is important that interventions are individualized, existing similarities in the symptoms and treatment of patients with CMT also merit therapeutic consideration. In the next sections, we provide an overview of these common areas and considerations for the care of people with CMT.
Mobility
It is important to note that there is large variability in the presentation of a person affected by CMT, even between individuals with the same type of CMT and/or within the same family. Outside of disease-specific outcome measures, your physical therapist has many tools, tests and outcome measures in his/her toolbox that can be used to monitor your functional mobility. A mobility assessment will be part of your initial physical therapy evaluation, and it will vary over the course of your life. Each time you walk through the door, your unique situation will be assessed for the most optimal plan of care.

The physical therapist will inquire how you are getting around and completing your everyday tasks. Are you experiencing excessive fatigue with your day-to-day activities? Are you experiencing falls? If so, what are the environments where your falls are taking place? Maintaining independence in mobility is a key area in which physical therapy intervention can be incredibly helpful.

When possible, using consistent standardized mobility tests can provide documentation of your symptom progression, as well as help determine the effectiveness of therapy interventions. Do not be surprised if you repeat the same tests that the therapist performed earlier in your care, as he/she is looking to compare the results. The tests the therapist chooses to complete will vary depending on your age and the state of your symptom progression. A video might be able to capture the way that you walk now and maybe demonstrate the effectiveness, or lack thereof, of an assistive device or orthotic. A therapist might also closely analyze the way that you walk, also known as gait analysis.

The physical therapist is hoping to capture where you are at this moment in time regarding your mobility. Again, his or her goal is to keep you moving in your daily life so that you are achieving your daily activities safely and successfully for as long as possible. Similar to the notes in the initial evaluation section, please communicate to your therapist your goals and preferences. His or her hope is to work with you to develop a plan to help you stay as active as possible for as long as possible.

Balance
Decreased balance and falls are often reported by individuals with CMT and have a significant impact on quality of life. Balance physiology involves the input of sensory information from the visual, vestibular (inner ear and balance) and somatosensory (sensation) systems. Information regarding the body’s position in space is gathered from the nerve sensors throughout the body and compared in the brain. The brain then activates the appropriate muscles to correct or maintain balance. These corrections are referred to as balance strategies.

In people affected by CMT, normal balance physiology is impaired. Somatosensory information may be limited due to the nerves’ inability to relay back an accurate signal, which may confuse the brain in interpreting your position in space. Weakness, specifically in these muscles, can decrease the ability to use these muscles to correct for small changes in balance, resulting in unsteadiness. Age-related changes in the vision and vestibular systems, as well as other health issues, affect balance. Different types of CMT and the stage of disease progression may result in variable presentations of balance impairment.

The assessment of balance is often guided by patient report, age and abilities. Assessment of sensation, strength and functional abilities, including gait and stair negotiation, are essential. An understanding of how your balance ability and confidence affects your participation in daily activities both inside and outside the home is also important.

Interventions to address balance issues are guided by the results of the assessment, as well as your personal goals. Interventions are often multi-dimensional and should be task-specific. Balance training may be effective. The use of orthotics (foot orthoses, lace up ankle supports, ankle foot orthotics) and assistive devices (walking sticks, canes, walkers and powered mobility devices) may be appropriate means of addressing balance limitations. It is also important to include patient education and fall management as part of the intervention.
Exercise and CMT

Everyone can benefit from regularly participating in activities that get the heart pumping and lungs working. This type of general exercise (cardiovascular training, also known as cardio) has been shown to have enormous benefits to our health, well-being, mood and ability to do what we want to do. It also can keep other illnesses at bay, such as heart disease, diabetes, high blood pressure and obesity. Studies of cardiovascular exercise in people with CMT have shown that fitness levels can be safely improved through regular exercise such as bike training. The same result may also be achieved with walking, swimming, rowing and other exercises, as long as the heart and lungs are working a little harder than normal. Be sure to check with your doctor before beginning any exercise plan and choose exercises that are safe in relation to your CMT expression.

Exercises to strengthen the muscles help to improve movement, balance and endurance. There is some indication that muscle-strengthening exercises can also help with the maintenance of blood sugar and bone density as we age. There have been a number of studies where weights have been used to strengthen the muscles at the tops of the legs and arms – the less affected muscles – in people with CMT. These studies showed that this type of exercise is safe and strengthens the muscles. The smaller muscles in the hands and the feet are more affected in patients with CMT. In children, a study in Australia found that foot and ankle muscles could be strengthened with weights. In adults, these muscles tend to be weaker and may not be able to lift a weight. No studies have looked at strengthening these weaker muscles in adults with CMT; therefore, it is not yet clear if using weights is safe or effective.

In general, we should all aim to sit less and break up long, sedentary periods of the day with movement. People with CMT have shown to be less active because it is more challenging to walk or participate in exercise with a disability. Sitting for long periods has been shown to relate to increased obesity in patients with CMT. Helpful strategies include setting reminders to move during the day and choosing fun activities that involve movement.

People who are new to exercise are often unsure how hard to work. A good way to self-monitor is to be aware of how much you are exerting yourself when exercising. Using a zero to 10 scale, you can imagine that you would have an exertion score of zero out of 10 while you are resting. If you are working so hard that you are close to collapse, that would be an exertion score of 10 out of 10. To derive benefit from exercise while not overdoing it, you can aim to exercise at a five or six out of 10. People who are more fit and accustomed to exercise may want to aim for an eight out of 10. Your doctor or an exercise specialist can help you determine what’s right for you.
Foot and Ankle Weakness
Exercise has undisputed health benefits and is potentially therapeutic for neuromuscular disorders such as CMT. But there is a risk of harm from overexertion and weakness. Progressive foot dorsiflexion weakness, also called foot drop, due to axon degeneration, is a debilitating problem in patients with CMT that can cause difficulty walking, painful deformities and reduced quality of life. To date, exercise trials for CMT have been limited to adults, proximal muscles and short-term outcomes (www.cmtausa.org/exercise-study-abstract).

In Australia, a team of rehabilitation professionals led by professor Joshua Burns, PhD, recently conducted a randomized controlled trial demonstrating that six months of progressive resistance exercise of ankle dorsiflexors was safe and effective in delaying strength loss in children and adolescents with CMT. This was the first randomized controlled trial to assess exercise therapy for pediatric CMT. It was found that six months of targeted moderate-intensity progressive resistance exercise of the foot dorsiflexors preserved long-term dorsiflexion strength without detrimental effect on the muscle size or shape and without producing other signs of overwork weakness. This trial provides evidence that the concept of progressive resistance exercise is safe and effective, and based on other studies in adults of similar muscle groups, suggests that progressive resistance exercise will also be safe and effective for other muscle groups under appropriate guidance and supervision.

STRATEGIES TO DISCUSS WITH YOUR PHYSICAL THERAPIST
Below is a summary of the exercise training program discussed for implementation in clinical practice. The exercise cuff used in the study was purposely built based on commonly available adjustable ankle weights that attached to the foot, rather than the ankle, with a heel strap to prevent slippage.

| DOSE | Both legs are exercised, one at a time, 3 days/week on non-consecutive days |
| WARM UP/COOL DOWN | 5-minute active warm up before training & a 5-minute cool down after training (consisting of slow walking) |
| INTENSITY | Initially 50% of 1Repetition Max—the weight a person can lift with maximum effort in a single repetition—for 2 weeks progressing to 70% of the 1Repetition Max |
| TIME | 25 minutes per session (and rest for an additional 30 minutes after the session) |
| EQUIPMENT | An adjustable weighted exercise cuff is required to perform these exercises |
| REPETITIONS | 8 repetitions in each set (each rep involves plantarflexion to dorsiflexion slowly through full range of motion, (concentric contraction), then returning to a plantarflexed position over a 4 second period (eccentric contraction) |
| SETS | Initially 2 sets for 2 weeks progressing to 3 sets for one foot before moving to the other foot |
| PATTERN | Rest intervals of 3 minutes between each set of repetitions |
| PROGRESSION | A graduated progression of greater resistance based on 1Repetition Max assessed every two weeks under supervision |
| SUPERVISION | Exercise with a physiotherapist, exercise physiologist, podiatrist or athletic trainer to monitor progression and safety |
| GUIDANCE | Exercise training program video on Lancet TV (see link below) |

Please find additional details at:
www.cmtausa.org/resistance-exercise-children

Video guide of the exercise training program which appears on Lancet TV:
www.cmtausa.org/exercise-training-program

Stories in the media about the trial:
www.cmtausa.org/effective-treatment-children
**Occupational Therapy**

**Goals of Occupational Therapy**
The initial evaluation will provide your OT with insight into your specific challenges with activities of daily living (ADL). Interventions and education to help maximize your independence and engagement in meaningful activities can be tailored to meet your individual needs. The plan of care will outline exercises and activities, adaptations or assistive devices, modification to the environment, splinting options and guidance/education designed to achieve these goals. Your plan of care may include programs for stretching and exercising, recommendations for adaptive equipment and potential home modifications, ergonomic solutions, upper extremity splinting, tremor reducing techniques, pain relieving modalities and energy conservation techniques. OTs use both remedial methods – the process of restoring or maintaining function – and compensatory methods – strategies or equipment that compensate for a deficit – of intervention.

**Hand Function**
CMT can result in sensorimotor impairments – involving both sensory and motor functions – in the upper extremities that present as early as the first decade of life. Typically, the distribution of impairment follows a distal (hands) to proximal (shoulders) pattern, with the hands being most impaired and the proximal muscles relatively spared, although more severe cases or those further along in their disease process may present with proximal weakness as well. Sensory disturbances in CMT can result in burning, tingling pain, numbness and loss of sensibility, which is the ability to detect size, shape, texture and temperature in the hands. Additionally, decreased proprioception, which is the ability to perceive the position of the hand/fingers in space along with movement, speed and excursion, can affect hand use during tasks.

Motor impairment in CMT can limit active movement of the hand, thumb and fingers, cause fatigue, result in decreased muscle strength and lead to progressive muscle wasting or loss of muscle bulk and hand contracture/deformity. The intrinsic muscles of the hand, which are small muscles within the hand itself that help with refined finger movement, tend to be affected the most. This can result in decreased dexterity and the inability to complete manipulative tasks. Extrinsic muscle weakness, which involves the larger, forearm-based muscles responsible for opening and closing of the fingers and movement of the wrist, can limit forceful grasp and movement of the wrist. Muscle cramping of the hands/forearms, cold intolerance resulting in stiff fingers and tremor of the hands can also be present in people with CMT and contribute to ADL limitations.

The ability to interpret peripheral sensory stimuli, carry out appropriate motoric responses and continually perform this cycle of sensorimotor adaptation throughout a task is essential for efficient and effective task completion. Due to the sensorimotor impairments discussed above, the completion of ADL tasks can be quite challenging. Tasks requiring bimanual (both hands) manipulation such as buttoning clothing, fastening zippers, managing jewelry and tying shoes tend to be the most challenging. These tasks require a steady hand, the ability to interpret important sensory stimuli and the use of the fingers to reposition, manipulate and accurately release small objects with the dominant hand while the non-dominant hand serves as a stabilizing force or functional assist. Other tasks such as opening jars, removing container lids and tearing open food packaging can be challenging due to muscle weakness. Decreased thumb mobility can limit the ability to use a pincer grasp, which uses the thumb to index finger tip, stabilize objects against the fingers for grasp like hold a water bottle and complete manipulative tasks. As a result, lifting small things such as coins, food, writing implements, etc., off a table surface can be very difficult. Limited thumb mobility can also affect the use of the thumb for text messaging, writing, stabilization of the computer mouse and joystick operation for gaming.

**STRATEGIES TO DISCUSS WITH YOUR OCCUPATIONAL THERAPIST**

**STRETCHING:** Maintaining joint integrity is crucial for preventing joint contractures and soft tissue shortening. Daily stretching may help with hand cramping as well as reduce pain and joint deformity. Remember, a stretch should never be painful!

- **Wrists**
  Flexion/extension range of motion stretches prevent stiffness and tightness.

- **Fingers**
  Stretching may help prevent proximal interphalangeal (PIP) flexion patterns and thumb stiffness due to muscle imbalance.
**STRENGTHENING:** Resistance training for hand and wrist muscles may help prevent muscle atrophy, as well as maintain hand strength for those with CMT. It may also improve strength for those with mild to moderate weakness. It is essential to understand the difference between intrinsic and extrinsic muscles and to target their strengthening appropriately.

**Extrinsic hand muscles**

Resistive movement activities utilizing gross grasp (making a fist), wrist flexion and extension (bending up and down) and pronation/supination (palm up/down) can help strengthen the hands.

**Intrinsic hand muscles**

Resistive movement activities utilizing the various pinch patterns (lateral, palmar, and pincher) abduction (spreading apart and together) of the fingers and an intrinsic plus grip pattern (MP joints flex while keeping IP joints fully extended) can strengthen the hands.

**Hand Function**

Activities and exercises that engage hands in coordination, fine-motor and manipulation practice may help increase efficiency and carry over into ADL performance.

**SPLINTING:** Your OT will determine if a splint is necessary to promote increased hand function and engagement in ADLs or to provide a stretch to prevent/convert a hand deformity. Below are some helpful splints for those with CMT.

**Tools such as Theraputty, clothes pins, snack clips, scissors, rubber bands, bubble wrap, sponges, rubber bottles can be used to provide resistance for the aforementioned movement patterns.**

**Stringing beads, lacings, needlepoint, etc., help with fine motor skills. Using Chinese medicine balls or golf balls, rotate the two balls in your hand clockwise and counterclockwise to help work on rotational movement of the thumb and coordination of side to side finger movements. Having small coins or dice in your palm and bringing them to your fingertips one at a time, placing them on a table top or into a piggy bank helps develop in hand manipulation skills.**

**Activities like sign language, playing the piano, flute and other musical instruments help work on coordination and isolated finger movements. Games like Nudge, building with blocks and cooking tasks (measuring/pouring) are all good activities to work on isolated finger movements.**
Managing Fatigue

Fatigue is a common symptom for people with CMT, but the word “fatigue” can be used to describe different experiences. There is a type of fatigue related to being active when the muscles get tired when a task feels like hard work. There is another type of fatigue that is a more general, which translates to overwhelming tiredness that is not always related to what you are doing. It is also more common for people with CMT to have sleep disorders, such as obstructive sleep apnea or restless legs syndrome. Poor quality sleep can make you drowsy and prone to nodding off during the day.

Fatigue related to activity is often resolved after rest or a good night’s sleep. This type of fatigue can respond to general exercise and strengthening exercises to increase muscle endurance. The more overwhelming, non-activity related fatigue does not always respond to rest. Some people report having a good night’s sleep but still waking up exhausted. Fatigue management programs run by occupational therapists can be particularly helpful for this type of fatigue. The focus is to explore triggers, strategies and energy-saving tactics to manage the symptoms. Gentle, graded exercise can also help, and some studies have reported that people feel more refreshed when exercising.

Obstructive sleep apnea can be characterized by excessive snoring, headaches and daytime sleepiness. If you think that this could be contributing to your fatigue, speak to your neurologist because there may be medical treatments that can help. This is also true for restless legs.
Studies of pain experienced by people with CMT have suggested that there are different causes of pain. One type is musculoskeletal pain, meaning the source of pain is the joints or soft tissues. This may be due to repeated injuries, like ankle sprains, or joint deformity and arthritis. This pain is often described as an ache or a deep pain. This type of pain is very common and is a secondary symptom of CMT because it is not directly due to the neuropathy. The other type of pain is less common and tends to more frequently affect people with Type 2 CMT because the smaller nerve fibers are affected. This is called neuropathic pain where the small pain nerve fibers are damaged and cause shooting or burning pain.

Distinguishing between the two can be difficult, but musculoskeletal pain tends to relate to movement and/or bearing weight through the joints. Neuropathic pain tends to be random and unrelated to movement. Some people find neuropathic pain is much worse at night.

Musculoskeletal pain can be relieved by using physical approaches to re-aligning or un-weighting joints. This could be accomplished by using stretching exercises, splints or braces (to re-align or protect a joint), or through surgery if there is fixed deformity. Seeing a physical therapist, orthotist or podiatrist to learn of existing options may be beneficial. Neuropathic pain can respond well to certain medications. Your neurologist can advise on this subject.

Summary
Hopefully, this information will allow you to prepare for and make the most of your consultation with a physical therapist and/or an occupational therapist. Please remember that CMT is a condition that most therapists do not routinely encounter. The therapist you are working with will appreciate any information you can share about CMT and how it affects you. Please share this booklet with your therapist and encourage him or her to seek additional information as needed.
Established in 1983, the CMTA is the leading source of information about CMT for more than 30,000 patients and families, supportive friends and medical professionals.

The CMTA offers a variety of educational materials and conferences, coordinates more than 70 CMTA branches nationwide, provides physician referrals and works closely with the clinical and research communities. The CMTA is also the leading financial sponsor of research within the CMT community, and it is the only CMT-specific patient advocacy organization in the United States strategically aligned with the National Institutes of Health Rare Disease Clinical Research Network (RDCRN).

Our vision of a world without CMT is taking shape! CMTA-funded researchers have identified promising drug candidates for CMT1A. One of them has not only stopped progression of the disease, but also showed improvement of some symptoms. The promise of symptom alleviation and the possibility of symptom reversal is real.

We invite you to be a part of the CMTA family and join us in our vision of a world without CMT.

www.cmtausa.org