Charcot-Marie-Tooth
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Charcot-Marie-Tooth

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Objectives

Objectives - Participants will acquire skills to (1) diagnose CMT based on common symptomology; (2) discuss work-up and surgical treatment options for the very young to adults; (3) identify the potential benefits of occupational and physical therapies and bracing to help normalize life for those diagnosed and their care-givers.

Target Audience:
• Neurologists, physical medicine and rehabilitation and other physicians interested in neuromuscular and electrodiagnostic medicine
• Health care professionals involved in the management of patients with neuromuscular diseases
• Researchers who are actively involved in the neuromuscular and/or electrodiagnostic research

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Pediatric Orthopaedic Surgery and the Hereditary Motor Sensory Neuropathies

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INTRODUCTION

The hereditary motor sensory neuropathies (HMSNs) are comprised of numerous diseases that result in abnormal peripheral nerve function. The most common is Charcot-Marie-Tooth (CMT) disease. These disorders have orthopedic problems and present with weakness, gait abnormalities, sensory loss, and progressive deformities. As with any disease, an understanding of the natural history is important to determine effective treatment. Because most of the HMSNs have only now been delineated by specific genetic markers, the orthopedic literature regarding diagnosis and treatment of specific HMSNs is scant and primarily descriptive, with few articles containing meaningful numbers of patients. Because CMT1a is the most common type of HMSN, more information has been published about it than the other diseases. Thus, CMT1a will be the primary focus of this discussion. However, many of the orthopedic manifestations are similar throughout this family of diseases, and many of the lessons from CMT1a can be extrapolated (out of necessity) to the less common forms of HMSN.

Early Recognition of Disease

Because HMSNs are chronic and progressive neuropathies, patients will experience a progression of symptoms. Initially, children may not complain of any noticeable weakness or deformity. Pes valgus and lack of reflexes may be the earliest signs. However, it should be remembered that pes valgus is a normal variant in the general population. The parents may also notice an ungainly gait and a lack of coordination in their child. Again, these are very nonspecific findings.

As peripheral weakness progresses, the child will gradually develop the more commonly recognized signs of CMT, including intrinsic wasting of the hands, cavus foot deformities, and claw toes. The gait abnormality becomes more obvious, often with development of a drop foot resulting in a “steppage” type gait. Accentuation of pelvic tilt to assist in clearance of the foot has been described as a “marionette” gait. Equinus contractures may develop as the gastrocnemius muscle overcomes the tibialis anterior and accessory ankle dorsiflexion muscles. Increased use of the accessory dorsiflexors of the foot including the extensor hallucis longus (EHL) and extensor digitorum longus (EDL) result in the development of claw toes. The first metatarsal drops, increasing heel varus and cavus due to a tripod effect. Ankle instability with frequent sprains is a common complaint, as well as the development of metatarsalgia and other foot pain. Eventually, as the cavus deformity becomes fixed, the patient will develop callosities under the first and fifth metatarsal heads and often over the base of the fifth metatarsal as well.

Additional orthopaedic problems encountered in CMT population include the development of scoliosis and hip dysplasia. The exact pathway by which these problems develop is still unclear. Finally, children with CMT often develop intrinsic weakness of the hands with progressive upper extremity dysfunction and problems with activities of daily living. These upper extremity issues are beyond the scope of this discussion.

FOOT DEFORMITIES IN CHILDREN: PREVENTION AND TREATMENT

While cavus is considered a sentinel manifestation of disease in children with CMT, some children will have pes valgus and some
will have no foot deformity at all. Limited ankle dorsiflexion, related to Achilles tendon tightness, may be one of the earliest signs. However, over time pes cavus becomes the dominant deformity. Weakness in dorsiflexion resulting in the tight Achilles tendon, accompanied by over pull of the EHL and EDL, results in clawing of the toes and accentuation of the plantar arch as previously discussed. This eventually becomes stiff and a fixed cavovarus deformity results.

Once the hindfoot is stiff in varus, correction of the deformity becomes very difficult and triple arthrodesis of the hindfoot to correct this deformity may be the only option to obtain a plantigrade foot. While many patients are happy with the results of triple arthrodesis, there is evidence that its benefits in CMT patients are limited. Eventual degeneration of the surrounding joints of the foot and ankle often result. Thus, triple arthrodesis should be reserved for those feet that have severe and rigid deformities (better straight and stiff than deformed and stiff), and every effort should be made to avoid progression to this point.

**Stretching, Splinting, and Strengthening**

Given the problems with triple arthrodesis noted above, the goal of treatment in children with CMT is to provide a stable, flexible, plantigrade, pain-free foot that lasts a lifetime and avoids the need for fusion procedures. While there is little evidence of the longterm benefits of stretching, night splinting, and strengthening exercises, there is some published literature supporting stretching and strengthening. Importantly, there is little risk to these interventions. Parents often wish to intervene as early as possible, and stretching, splinting, and strengthening are simple, safe modalities that can be employed by the practitioner.

**Gait Abnormalities and Bracing**

Associated with the weakness in dorsiflexion are gait abnormalities. Most often, a steppage gait is seen where the patient hyperflexes their hip to avoid catching the toe during the swing phase. Further weakness of the hip flexors may result in overcompensation of pelvic tilt resulting in the “marionette” gait mentioned above. Simple treatment options include strengthening of the tibialis anterior and the use of an ankle-foot orthosis (AFO) to prevent tripping. Different types of braces are available, including the leaf-spring AFO which keeps the foot slightly dorsiflexed but allows slight plantar flexion on heel-strike due to the cutback of material posterior to the ankle joint (Fig. 1). More complicated braces with hinged ankles are also available but provide the same function.

It is important to remember that the function of a static brace is to hold a joint in a particular position. If the ankle cannot be brought to a neutral position, static bracing will not be possible. Once an equinus contracture has developed, dynamic night bracing may assist in stretching the Achilles tendon. Slight equinus contracture of only a few degrees is well tolerated, particularly since most shoes have a built in angle of plantar flexion. As the equinus worsens, lengthening of the Achilles tendon may be needed to get the foot into a neutral position. Over-lengthening of the Achilles tendon can result in significant weakness of the calf muscles and a calcaneus deformity, with decreased toe off and decreased stride length. In addition, a calcaneus deformity is difficult to brace and may result in significant pain and decreased endurance. Equinus is always better tolerated than calcaneus, so care should be taken never to over-lengthen the Achilles tendon.

**Tendon Transfers and Osteotomies**

As weakness and deformity progress, efforts should be made to balance the motors of the foot. Tendon transfers can improve this balance by removing deforming forces and providing more power to weak muscles. There are a number of preconditions to successfully transfer a tendon. Some that are of particular importance to this population include:

- The muscle to be transferred must be expendable.
- The muscle must have sufficient strength (grade 4 or greater).
- In its transferred position the muscle must have action over a mobile joint.
- It is best to transfer in-faze muscles, if possible.
- The muscle must have adequate excursion to perform the intended function.

In order to determine if the hindfoot is mobile, the Coleman block test is utilized. Plantar flexion of the first metatarsal results in a varus tilt of the heel when the foot is in the standing position (Fig. 2). To establish the presence of mobility of the hindfoot, the patient stands on a block allowing the first metatarsal head to drop off the side of the block. If the heel moves into a valgus position, the hindfoot is flexible. If the hindfoot remains in varus, the deformity is rigid and transfers with osteotomies are likely to fail.

Muscles that frequently can be transferred in patients with CMT include the EHL to the first metatarsal (often in conjunction with a first interphalangeal fusion, known as a Jones transfer), the EDL of the lesser toes to the metatarsals (a Girdlestone-Taylor transfer), or the EDL of the lesser toes to the dorsum of the foot (a Hibbs transfer). These transfers improve strength by decreasing wasted motion across the interphalangeal joints as well as removing a deforming force. In addition, one can transfer the peroneus longus to the peroneus brevis to increase eversion strength and decrease...
plantar flexion of the first ray. The decision to transfer any of these muscles is dependant on those preconditions noted above and the severity of the deformity.

Osteotomies often are included when transferring tendons in order to realign the foot. The two most frequent osteotomies are the Priddie osteotomy (also known as the Koutsogiannis osteotomy), in which the heel is pushed into valgus in order to move the point of heel strike laterally, and a first metatarsal dorsiflexion osteotomy to lift the great toe (Fig. 3). Standing anteroposterior and lateral radiographs of the foot give a fair evaluation of the cavus deformity, but they do not show the position of the heel in stance. The Cobey view is very useful in that it allows determination of the point of contact of the heel in relation to the mechanical axis of the tibia (Fig. 4).

**Triple Arthrodesis**

Finally, in those feet with a rigid hindfoot, triple arthrodesis may be the only option. Wedges of bone from the hindfoot and midfoot may need to be removed in order to realign the foot into a plantigrade position. Early failures of triple arthrodesis are usually the result of under correction. In order to dorsiflex the midfoot, a wedge of bone may need to be removed from the midfoot. Preoperative planning is important and it should be noted that this wedge is a trapezoid. The apex of any wedge taken from the midfoot must be at the plantar surface, as this is the point of rotation, and not at the inferior surface of the bone. Again, while the results of triple arthrodesis are of concern in longterm outcome studies, the operation may be the only option to obtain a plantigrade foot.

**Scoliosis**

Scoliosis appears to be more common in the HMSN population than was initially thought. Scoliosis has been documented in 15-37% of patients with CMT, and it has been reported in other HMSNs. The most severe deformities seem to occur in Dejerine-Sottas syndrome, with 100% of patients affected in one study. Of CMT patients with scoliosis, 71% of curves were found to progress. Unlike adolescent idiopathic scoliosis, which is usually hypokyphotic, scoliosis associated with CMT may be kyphscoliotic. Most curves do not appear to respond to brace treatment but not all require surgical intervention.

The surgical approach for scoliosis associated with CMT is similar to that of the adolescent idiopathic scoliosis population (Fig. 5), with good results reported by Karol and Elerson. They noted that they could only utilize intraoperative somatosensory evoked potentials in 25% of their surgical patients. However, there were no reported neurological deficits postoperatively.

**Hip Dysplasia**

Hip dysplasia also has been associated with CMT. Walker and colleagues noted hip abnormalities in 35% of their patients. Chan and colleagues assumed that the hips of CMT children were normal at birth and that dysplasia develops later. They recommend a standing anteroposterior radiograph of the hip every 2 years, and any infant with known CMT should undergo an ultrasound. The
cause of the hip dysplasia in CMT is unknown but likely is the result of muscle imbalance around the hip. Because of this, it is unclear if nonsurgical means (bracing) can alter the natural history of hip dysplasia in children with CMT. The dysplasia seems to develop later and insidiously. Patients with hip dysplasia may remain asymptomatic for extended periods of time. Due to the variability in disease severity in patients with CMT, age at which hip dysplasia develops also is variable.

Generally, surgical intervention to address the acetabular dysplasia by redirection osteotomy is recommended, with the subsequent addition of intervention on the femoral side if necessary. Proximal femoral varus osteotomy may result in a worsening of gait, so is best performed if the acetabular procedure fails to address all of the dysplasia. Unlike scoliosis treatment in CMT, neurologic dysfunction after hip surgery has been reported, so care should be taken not to stretch the peripheral nerves. Chan and colleagues stresses the importance of these patients walking as soon as possible after surgery.

**CONCLUSIONS**

The HMSNs are associated with a number of orthopaedic problems including foot deformity, spinal deformity, and hip dysplasia. Young patients may present with flat feet and toe walking. Achilles tendon stretching, dorsiflexor strengthening, and bracing may delay the onset of cavus deformity in young patients. Early surgery in cavus feet, including tendon transfer and osteotomy to rebalance the foot and correct primary deformity, may prevent patients from later requiring triple arthrodesis.

Patients with HMSN should be routinely screened for scoliosis by physical examination and for hip dysplasia by radiographs. Bracing may not be successful for patients with scoliosis, but surgery seems to be safe and effective if required. Hip dysplasia may be silent and develop late in childhood and adolescence. Surgery to address the acetabular dysplasia should be performed first, and care should be taken to avoid stretch injury to peripheral nerves.

**REFERENCES**


**Figure 5.** (A) A 15-year-old boy with Charcot-Marie-Tooth disease and a 65 degree thoracic curve. (B) Surgical correction with posterior instrumentation and fusion was uneventful.
INTRODUCTION

Charcot-Marie-Tooth (CMT) disease, also known as hereditary motor and sensory neuropathy (HMSN), is a slow, progressive, peripheral nerve disorder, which, in its most prevalent form (CMT1a, 70% of all CMT type 1 cases), results in muscle atrophy, weakness, and decreased nerve conduction velocity. CMT is the most common inherited peripheral sensorimotor neuropathy, affecting 1 in 2,500 persons. The expression of the disease ranges from mild-to-severe, usually with slow progression. Patients with mild symptoms experience varying degrees of distal limb muscle weakness. The weakness is due to a combination of factors, including axonal death due to associated demyelination of peripheral nerves, deconditioning, and disuse atrophy. A common impairment seen in individuals with CMT is decreased strength in intrinsic foot and lower extremity muscles, especially in the dorsiflexor muscles. Weakness also is a concern in the upper extremities, usually to a lesser extent. This lower extremity weakness, combined with the disease process of CMT, leads to problems, such as an increased propensity for ankle sprain, abnormal gait, abnormal posturing, and discomfort in the lower extremities. Those with more severe complications suffer foot and hand deformities, repeated ankle sprains, intense pain, chronic fatigue, extremely limited ambulatory ability, and distal amputation. Additionally, the psychosocial impact is comparable to patients with stroke and similar impairments. Contemporary Treatment and Management of Charcot-Marie-Tooth Disease: Embracing the “Exercise Is Medicine™” Model

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CMT does not appear more frequently in any particular geographical region, age group, race, gender, or ethnic group. It was first described in the late 1880s by three neurologists: Jean Martin Charcot, Pierre Marie, and Howard Henry Tooth. During the 50 years following its initial description, many variations of CMT were discovered and described. Yet, it was not until the 1970s and 1980s that scientific and clinical clarity further described and classified CMT. The use of electrophysiology and genetics helped delineate the classifications, as patients with CMT are now grouped by either the results of nerve conduction velocity studies or genetic tests. Today, the most conclusive and acceptable confirmation for the presence of CMT disease is accomplished with genetic testing. This applies, however, only to those CMT types and subtypes which currently may be evaluated by genetic assay.

No cure exists for CMT, and treatment for CMT traditionally has been focused on limiting impairments caused by the disease process. Such intervention historically has included surgical orthoses (tendon transfers) and prescription of orthotic devices. The clinical literature indicates that ankle-foot orthoses (AFOs) have been found efficacious in improving physical performance and decreasing the rate of perceived exertion in patients with CMT. Gait training has been found to decrease the amount of energy patients expend while walking, as well as improve stability and strength. Resistance training has been found efficacious in improving strength and activity of daily living (ADL) performance in adults with CMT.
Aerobic training has been used to decrease the fatigability and improve the exercise capacity of patients with CMT and other neuromuscular disorders.\textsuperscript{11,17} The efficacy of aerobic training over resistance training in CMT patients is somewhat equivocal, primarily because very few systematic basic science investigations exist and, translationally, no clinical trial has directly compared these two exercise modes. Other investigations have examined the use of drugs (e.g., neurotrophin, and prednisone) and nutritional supplements (e.g., ascorbic acid, curcumin, and creatine monohydrate); effectiveness was not demonstrated, or, at best, was uncertain.\textsuperscript{5,24} Though no clinical intervention has been standardized to treat CMT, the literature indicates that regular exercise and activity training may, presently, provide the best alternative to manage some of the symptomatology associated with CMT.\textsuperscript{1,7,8,11,16,17,18,19,20,22,28,36}

**LITERATURE SYNOPSIS: THE CASE FOR EXERCISE AND ACTIVITY TRAINING**

Several forms of intervention have been examined in the treatment of CMT; however, none have proven reliably effective. This includes drugs (e.g., analeptic agents, progesterone antagonists, and neurotrophic factors), nutritional supplements (e.g., ascorbic acid, curcumin, and creatine monohydrate), and surgical procedures (e.g., osteotomy, fasciotomy, tendon transfer, tendon release).\textsuperscript{5,9,24} One type of intervention, exercise, has, however, shown some reliable effectiveness—across studies—in the treatment and management of CMT.\textsuperscript{17,18,36}

Though not extensively examined in clinical science, the use of regular exercise and physiotherapy as a treatment for CMT has demonstrated some encouraging results. CMT patients typically have not been referred for exercise or physiotherapy intervention, but the available data indicate that resistance exercise training improves strength, functional outcomes, and morphologic measures in these patients.\textsuperscript{1,7,8,16,19,20,22,28} A smaller number of studies have demonstrated that different forms of aerobic training may improve aerobic capacity, functional ability, and risk exposure to comorbid disease in CMT patients.\textsuperscript{7,8,11}

Only a dozen or so studies have examined the use of exercise as a treatment for CMT. Most of these have investigated the use of resistance training in patients with CMT1a. An early study investigating the effects of resistance training established a link between muscular strength and functional ability in patients with CMT.\textsuperscript{19} Since then, it has been determined that the training-induced increase in strength is significantly related to improved ADL performance.\textsuperscript{7,8,18}

Overall, the literature reveals that resistance training protocols, of at least 12 weeks duration, used in adult CMT populations have proven the most effective. This includes reported beneficial changes in strength, power, functional ability, aerobic capacity, body composition, muscle fiber size, and muscle protein composition.\textsuperscript{7,8,18,24} One such investigation examined the effects of low-to-moderate resistance training on aerobic capacity, body composition, muscle fiber type and size, strength, and timed ADLs in 20 CMT patients.\textsuperscript{7} Beneficial changes in strength, muscle fiber size and protein composition were observed. Additionally, these muscle adaptations strongly correlated with improved ADLs.\textsuperscript{7,8}

Though the resistance training did not produce statistically significant improvement in body composition for CMT patients, a clinical change in increased lean mass of 1.5 kg was reported; this was consistent with previous findings of increased lean mass for age-matched healthy persons engaged in similar resistance training programs.\textsuperscript{2}

Two case-reports have investigated the use of resistance training in children with CMT. A 12-week lower extremity, high-intensity resistance exercise program (up to 80% of one-repetition maximal strength) for a 15-year-old girl with CMT resulted in clinical improvements in plantar flexion and dorsiflexion strength, jumping power, and walking ability.\textsuperscript{5} No adverse events or deleterious effects of the trained and associated muscle groups were reported.\textsuperscript{9} Another case study, examining the effects of combined moderate-to-moderately-high intensity, resistance, and aerobic training over 1 year in a 16-year-old girl, reported clinical improvements in blood lipids, lean mass, bone mineral density, upper and lower body strength, and several balance measures. The latter included specific beneficial changes in rhythmic weight shift, weight-bearing, limits of stability, and sensory interaction on balance (Chetlin, personal communication, 2007. This apparently is the only research that has examined both efferent [motor] and afferent [sensory] effects of exercise training in any patients with CMT).

The literature indicates that resistance training studies involving CMT patients have used variable amounts of intensity. Though clinical consensus has favored a low-to-moderate intensity approach, based on reported results, resistance training at higher intensities has produced some equivocal results, ranging from no indication of muscle damage to self-reported overuse.\textsuperscript{2,18,19,22} Though no study has evaluated the effects of traditional (i.e., continuous) aerobic training or the concurrent effects of resistance and aerobic training in CMT exclusively, when patients engaged in aerobic-type resistance training for 12 weeks, their type I (i.e., oxidative) fibers hypertrophied and their type I myosin heavy chain proteins increased.\textsuperscript{7,28} Timed motor performance of functional ability (e.g., chair rise, supine rise, and stair-stepping) and total time of exercise also improved in these patients. A novel finding of this study was that CMT patients are at increased risk for cardiovascular disease due to high body fat percentage and high body mass index (BMI), thus classifying them as obese.\textsuperscript{2,7} Seventeen percent of these patients had type 2 diabetes and patients exhibited low exercise capacity, achieving only 5.7 times above resting metabolic equivalents (METS) during graded exercise testing. A peak exercise capacity of 5.7 METS doubles the risk of all-cause mortality compared to individuals with peak capacities of 8 METS or greater.\textsuperscript{23}

Interestingly, a 24-week program of interval training (i.e., a form of aerobic conditioning, characterized by short, high-intensity bouts interspersed with longer, low-intensity training stanzas) on a bicycle ergometer improved maximal power, lower body isokinetic strength, and timed-motor functional ability in patients with CMT1a and CMT2.\textsuperscript{11} Two other important findings were gleaned from this study. First, patients demonstrated the ability to intermittently train at higher intensities (i.e., 1-min bouts at 80%
of maximal power and 70-90% of maximum heart rate) without adverse effects. Previous investigations had determined that high-intensity training—in the form of resistance exercise—may produce overuse/overreach symptoms, while low-to-moderate intensity training would likely avoid such complications. Thus, there might be certain CMT patients who may, without contraindication, exercise at higher levels of exertion.

Second, patients improved their maximal oxygen consumption by one MET over the 24-week training period. It is well established that regular aerobic training improves exercise capacity and even a small increase in exercise capacity (i.e., 1 MET) is associated with an 11% reduction in all-cause mortality in both young and old populations. A 1 MET increase in peak exercise capacity also translates into a 5.4% decrease in healthcare costs associated with a sedentary lifestyle. Considering that combined direct costs of physical inactivity and obesity account for approximately 9.4% of healthcare expenditures in the United States, even small improvements in exercise capacity may translate into substantial healthcare cost reduction in this population. Given that many CMT patients have high BMIs, poor body composition, and poor exercise tolerance, improvements in exercise capacity may reduce risks for coronary artery disease, other comorbidities, and all-cause mortality. It would also reduce the cost to treat this group of patients from the effects of a sedentary lifestyle.

Despite the established benefits of exercise shown in several studies, the specific question of whether or not individual CMT patients can exercise safely should be addressed. The literature indicates that CMT patients may engage in regular exercise, provided contraindications to exercise are not present. These would include known cardiopulmonary disease, poorly controlled diabetes or hypertension, severe orthopedic limitations, and debilitating weakness (i.e., strength levels <15% of normal values). The latter two are not pure contraindications, because many modes of exercise are likely available to avoid orthopedic complications, and the amount of strength present appears to be related to the expected benefit from the exercise itself. That is, very weak patients may not derive the same magnitude of improvement as those patients who possess more initial strength prior to the commencement of an exercise routine. Additionally, muscle strength appears to be inversely correlated to the histopathology of muscle fibers. Therefore, CMT patients, who exhibit the least amount of disruption of their muscle fibers, display greater amounts of strength and may reap greater benefit from an exercise training program.

Given that there currently is neither a cure for CMT, nor any standardized treatment protocol to manage the disease, the literature supports the use of regular exercise to improve strength, endurance, and daily function in most CMT patients, as well as reduce their risk of incurring comorbid disease owing to a sedentary lifestyle. Today, allied health practitioners have at their disposal the means to recommend exercise as a scientifically-based clinical intervention to treat patients with chronic disease, including CMT. The goals of clinically-prescribed exercise should focus on improving health, wellness, and quality-of-life through regular physical activity and healthy behaviors.

In 2010, the American College of Sports Medicine hosted the First World Congress for the “Exercise Is Medicine™” (EIM) initiative in Baltimore, Maryland. Together with the recommendations provided by the Physical Activity Guidelines for Americans, published by the United States Department of Health and Human Services, EIM actively promotes these scientifically-based physical activity guidelines to facilitate the important health benefits of exercise and activity for all Americans, including persons with chronic disabilities, such as CMT. The clinical aim of EIM is to incorporate activity assessment and exercise prescription, when appropriate, as standard clinical operating procedure in the prevention and treatment of disease. Other organizations, such as the American Medical Association, the United States Department of Health and Human Services, and the Office of the United States Surgeon General, all agree that children and adults with chronic disease and disability (including CMT), whom are capable, should engage in regular forms of activity and exercise.

Children with CMT should participate in aerobic, muscle-strengthening, bone-strengthening, and balance exercises and activities. Collectively, this should amount to about 1 hour of exercise/activity per day. This should include at least 3 days of moderately vigorous variable activity that is fun, age-appropriate, and not contraindicated for the individual child’s medical circumstances. Because CMT is both a motor and sensory neuropathy, it is important to recognize the individual limitations in order to minimize exposure to injury. In most cases, motor control and sensory awareness of the ankle and foot are impaired. Under these circumstances, activities involving running and jumping—or other activities where the feet intermittently or continuously leave the ground—may not be safe.

Alternative exercises and activities should be considered for children with CMT, including bicycle riding (e.g., tandem riding, with the child at the front of the bike and the parent/adult at the back of the bike), games of catching and throwing, swimming, resistance exercises (using the child’s own body weight, exercise bands, machines, and hand-held weights), “exer-games” (video exercise/activity games on the Wii, Playstation, or Xbox game consoles), and lower-intensity martial arts (e.g., tai chi). Additionally, children with CMT should be encouraged—as all children should—to participate in appropriate forms of play. If children have been prescribed orthopedic braces, these should be worn during exercise and activity where possible.

For children with CMT, regular activity, including play, contributes to normal, healthy physical and psychosocial development. Certain clinical evidence indicates that early intervention may attenuate the disease process, possibly contributing to higher levels of function and quality-of-life into adulthood. Additionally, children who adopt an active lifestyle are less likely to be exposed to the eventual dangers of inactivity, including obesity, heart disease, and type 2 diabetes.
Adults with CMT should also participate in various forms of aerobic, muscle-strengthening, bone-strengthening, and appropriate balance training. Ideally, it is recommended that adults with CMT should perform about 150 min of total aerobic exercise and activity per week. Such activity should be performed in moderate to moderately vigorous bouts of at least 10 min duration, spread throughout the week.\textsuperscript{32} Muscle and bone-strengthening exercises/activities—especially those involving simultaneous use of all major muscle groups—should be performed at least 2 days/week.\textsuperscript{32} In cases where significant motor and sensory impairment of the ankle and foot are present, exercises and activities where the feet leave the ground, either intermittently or continuously, should not be performed. Some examples of low-impact exercise/activities include riding or rowing exercise, swimming and water aerobics, vigorous gardening (with components like digging and lifting), household chores, resistance exercises (with bands, machines, dumbbells, and/or bodyweight resistance), and low-impact balance and flexibility training (e.g., yoga and tai chi).\textsuperscript{12} Though 150 min of total weekly exercise/activity is recommended, adult CMT patients should avoid inactivity; therefore, some physical activity is always better than none.\textsuperscript{32} Prescribed orthopedic braces should be worn during exercise and activity where possible.

For adults with CMT, regular exercise and activity may improve, or at least maintain, functional ability, independence, and higher quality of life. Like all older adults, an active lifestyle helps to maintain muscle mass and bone health, as well as reducing incidence of falling and avoiding diseases associated with inactivity.\textsuperscript{32}

\textbf{FUTURE RESEARCH}

The vast majority of externally-funded CMT research in the past 10-15 years has focused on the molecular basis of this heterogeneous group of genetic disorders. During this period, most of this research has used experimental animals, primarily mouse models, to identify a multitude of genes responsible for the diverse mutational mechanisms that cause CMT. To date, 36 loci and at least 24 genes have been implicated in CMT, elucidating mutation, duplication, and deletion phenomena involved in abnormal myelination, axonal transport, Schwann cell differentiation, signal transduction, mitochondrial function, endosomal translation, and DNA repair.\textsuperscript{27,30,31} These animal models have been critically important in identifying potential drugs and molecular therapies that may, one day, modify and normalize gene expression in the form of a cure.

The logical question that arises from this body of work is, "Where do we go from here?" Of course, the potential efficacy of drugs and gene therapies must continue to be evaluated at the molecular level using the described animal models, but also a fully translational and bi-directional paradigm (re: "bench-to-bedside-to-bench") must be developed that not only satisfies the scientific community but also creates optimism in the CMT community, including patients and their families. Furthermore, such a process must allow for refinement and adjustment of the exercise model as a more integrated understanding of the basic science, clinical evaluation, and patient feedback becomes available. There exists a palpable frustration amongst patients today that the scientific community has become so enamored with the molecular dialectic that direct intervention or treatment for patients currently has been relegated to a lower tier of importance (Chetlin, personal communications, 2007, 2011).

Given that the body of evidence that exercise and physical activity has produced promising results across studies and clinical populations, perhaps such regimens should be given careful consideration as part of a fully translational model in CMT research. Exercise has been used effectively in several clinical circumstances, including, for example, treatment for type 2 diabetes, cardiac rehabilitation, hypertension, cancer, orthopedic rehabilitation, and the neuromuscular syndrome associated with aging.\textsuperscript{26} Furthermore, appropriate exercise prescription has been shown to improve patient sensitivity to various drugs, thus demonstrating a permissive effect between regular exercise and specific pharmaceutical agents, such as cardiac (e.g., cardiac glycosides and nitroglycerin), antihypertensive (e.g., ACE inhibitors and calcium channel blockers), and hypoglycemic (e.g., insulin and sulfonylureas) medications.\textsuperscript{15,21}

The interventional “redundancy” associated with exercise—a demonstration of effectiveness across clinical populations—is not surprising. Acute and chronic physical activity is associated with systemic upregulation, including biochemical, hormonal, physiologic, and molecular responsiveness.\textsuperscript{2,14} These beneficial effects have been recognized, of course, in the EIM model, a near-universal clinical intervention endorsed by the American Medical Association, American College of Sports Medicine, and the Office of the Surgeon General.\textsuperscript{26} The spectrum of desirable exercise effects has been classically observed, for example, in the treatment of type 2 diabetes; a disease with multiple hallmarks, including diabetic neuropathy. Patients who exercise not only become more sensitive to their hypoglycemic drugs, but exercise alone is considered insulin-mimetic; most compliant patients, who adhere to their exercise prescription and dietary modifications, eventually do not require drugs to combat this metabolic disorder.\textsuperscript{33}

If exercise has demonstrated effectiveness in a multitude of animal and human models associated with multiple clinical conditions, then why has it not been more extensively examined in the treatment of other diseases, such as CMT? How may it be possible to know if exercise, combined with drugs or genetic therapies, may promote an additive effect greater than either the drug or exercise effects alone? The answers to these questions should serve as the foundation to include an additional arm to the present CMT research direction.

The mouse models are, of course, extremely important; these studies not only reveal the genes responsible for CMT, but they can help to identify those molecular and biological mechanisms resulting in pharmaceutical and gene therapies to treat this disease. These models should, however, be complemented by a fully translational component that includes aerobic, resistance, and combined exercise. This initially may take the form of rodents exercising on treadmills, dynamometers, or both—with and without the potentially effective drugs and gene therapies identified by way of the mouse models. Part of such a process would include identifying those outcomes of localization, distribution, quantification, and performance that could be carried over to human trials. This approach, or one similar to it, if
for CMT ultimately will be discovered.

personal hope—that one day, in the not-so-distant future, a cure for CMT will ultimately be discovered. However, they can personally identify with and the hope—a very real and personal hope—that one day, in the not-so-distant future, a cure for CMT will ultimately be discovered.

REFERENCES


INTRODUCTION

Charcot-Marie-Tooth (CMT) disease presents with a complicated combination of lower limb deformities, muscle weakness, and balance loss. It is important to take these under consideration when developing a treatment plan that involves prescribing lower limb orthoses. Although CMT is genetic, there usually are marked differences between affected family members. They can be affected at different ages and with varying severity of symptoms. For this reason, each individual that presents with CMT is unique and should be treated as such.

SYMPTOMS

CMT symptoms may include the following:

- **Foot drop:** This occurs when the forefoot drags and fails to clear the floor. The patient may compensate by exaggerated hip and knee flexion.\(^1\) This occurs due to lack of dorsiflexion control. This can be noted with a side view observation of the patient. The foot drops to the floor at the beginning of the stance phase and during swing phase.

- **Pes cavus deformity:** This is an unduly high arch, an exaggeration of the normal longitudinal arch of the foot.\(^2\)

- **Varus deformities:** These deformities are present when the angulation of the body part is toward the midline of the body (bent inward).\(^3\) Deformities occur at the subtalar joint. The patient walks on the lateral border of the foot. The subtalar alignment in varus is in the position of supination and adduction.

- **Valgus deformities:** These deformities are present when the angulation of the body part is away from the midline of the body (bent outward, twisted).\(^3\) Deformities occur at the subtalar joint. The patient walks on the medial border of the foot. The subtalar axis moves in the position of pronation and abduction. This is best observed from the front and back of the patient.

- **Muscle atrophy:** This is wasting of muscle tissue.

- **Balance loss:** This can be observed when the patient is constantly moving their feet or using excessive knee flexion in order to lower their center of gravity for stability. Factors that contribute to balance loss in CMT include loss of sensory feedback, muscular weakness, and neuropathy.

ANALYSIS OF PATHOLOGICAL GAIT

Systematic analysis is a valuable clinical tool for determining the nature and severity of the patient’s skeletal or neuromuscular deviations/deficiencies. This also helps assess the adequacy of orthoses and other aids intended to assist in achieving a more normal ambulation.\(^5\)

Many gait deviations are created by the neuromuscular patterns produced from CMT. These are a result of nerve loss, muscular weakness, and ligamentous laxity. This almost always results in a reduction of normal walking speed. Some of the more difficult gait deviations that occur include:

- Hip hiking (Fig. 1)
- Circumduction
- Internal or external limb rotation
- Abnormal walking base
- Lateral trunk bending
- Hyperextended knee
- Excessive knee flexion
- Insufficient toe off
DETERMINING THE CORRECT TYPE OF ORTHOSIS

Ankle-Foot Orthosis Versus Knee-Ankle-Foot Orthosis

Factors used when determining and prescribing a lower limb orthosis include age; overall strength; hand involvement; quadriceps strength; tibialis anterior strength; gastrocnemius strength; extent of damage to muscles, tendons, ligaments, bones, and joints; as well as balance.

Ankle-Foot Orthosis

An ankle-foot orthosis (AFO) is any orthotic device for the lower limb that encloses the ankle and foot and does not extend above the knee and is intended to prevent a foot from dropping due to inadequate dorsiflexion.

Knee-Ankle-Foot Orthosis

A knee-ankle-foot orthosis (KAFO) is any orthotic device for the lower limb that extends from above the knee to the ankle and foot. A KAFO would be used with a patient with a foot drop as well as a weak quadriceps that prevents the patient from being able to control knee flexion. Another consideration for a KAFO would be knee hyperextension greater than 15 degrees that cannot be controlled below the knee.

Variations of Ankle-Foot Orthoses and Knee-Ankle-Foot Orthoses

The current types of lower limb orthoses for foot drop include off the shelf models that come in small, medium, and large for both the left and right feet as well as custom made models that are fabricated from a mold of the patient’s lower limb that enables an exact fit with patient-specific corrections to the joints. The various mechanical types include posterior leaf spring, jointed, solid ankle, floor reaction, and energy-storing (Helios® type). The materials used include: metal, leather, thermoplastics, carbon fiber, and composites.

Selecting the Appropriate Device for the Charcot-Marie-Tooth Patient

Most CMT patients have similar symptoms, but their deformities and muscular weakness manifest in a myriad of ways. To determine the primary muscles that are affected due to the neuropathy, manual muscle testing to perform during a patient’s evaluation. This will help determine what type of brace would best serve the individual CMT patient.

FOOT DROP

Foot drop is a result of a neuropathy of the deep peroneal nerve which then does not send a signal to the tibialis anterior to dorsiflex the foot. Most people affected complain that when they walk their toes catch the ground and they trip and fall. Gastrocnemius weakness is also associated with this. Patients with gastrocnemius weakness cannot plantar flex, and this affects their gait. When walking, they cannot push off the ground to initiate the swing phase. Thus, the most common symptoms affecting walking seen in CMT patients are tibialis anterior weakness and gastrocnemius weakness. General muscle atrophy is also seen. The most common deformity seen among all CMT patients is pes cavus (Fig. 2).

The other two primary deformities seen in CMT patients are varus and valgus. Although there are no correlations of studies on this, it is the author’s experience that most patients with CMT1 fall into a varus angulation and most patients with CMT 2 fall into a valgus deformity.

BALANCE LOSS

The most common symptom experience by all CMT patients is balance loss. They rely on leaning on objects while standing. They always ask if there is a brace that can help them restore their balance; however, this is not possible. But, better balance can be achieved through proper bracing and corrections.

CMT patients also have a high degree of fatigue that sets in after walking for short periods. They have a balance loss combined with a foot drop and a possible varus or valgus deformity that is rotating.
the foot medially or laterally. This produces gait compensations in addition to a loss of energy, as they are using other muscles more strenuously in order to compensate.

GAIT COMPENSATIONS

The most common gait compensations seen in CMT patients include the following:

- **Hip hiking:** The patient elevates the hip on the affected side during the swing phase of gait to advance the limb and to clear the ground. This produces a marching or steppage gait.
- **Lateral trunk bending:** The patient moves from side-to-side in order to clear the ground. This can be caused by a wider walking base.
- **Circumduction:** The patient cannot clear the ground causing the limb to take a lateral semicircle path. This can be due to weak dorsiflexors.
- **Insufficient toe off:** The patient cannot get the toe off of the ground due to a weak tibialis anterior muscle. This also causes the gait compensation of hip hiking.
- **Abnormal walking base:** The patient develops an abnormal walking base due to general weakness of the lower limbs or a fear of falling. The space between heel centers is greater than 2-4 in.
- **Extensive knee flexion:** Many patients who do not have extensive knee hyperextension will go into knee flexion. When flexing the knees, they lower their center of gravity to the ground, which gives them better balance as well.
- **Hyperextended gait:** The patient’s knee extends fully and can go beyond the normal range. This occurs between heel strike and heel off. Possible causes are quadriceps weakness or ligamentous laxity.

ANKLE-FOOT OTHOSES: A PRODUCT OR A CLINICAL SERVICE?

Are AFOs a product or a clinical service? The answer is they are a combination of both. As a product, an AFO’s fabrication relies on proper evaluation of the patient, proper molding techniques, proper cast modifications, and proper mechanics that are put into the mold, all of which are responsible for the outcome of the finished AFO. And, the final AFO must be fabricated properly. It must fit well, be comfortable, help restore balance, and help produce a better gait. As a clinical service, an AFO must meet the four essential criteria that define an orthosis: it must be an appliance that will support, align, prevent, or correct deformities.6

Casting a Limb Mold for Maximum Correction

Before a custom AFO is made, a mold is taken of the patient’s lower limb. Deviations in the foot and ankle must be taken into careful consideration before the mold is taken. What must be corrected? What must be stabilized? If the patient’s foot has a high degree of valgus and/or midtarsal collapse where the foot deviates medially, that foot needs to be corrected as much as possible in order to prevent further deformity and to stabilize the foot and ankle. Additionally, this will help prevent further breakdown of the deviated foot in that plane of motion. As much manual correction as possible is desired for the deviated joint so that the patient can use the AFO to functionally control those corrections. Three-point corrective systems and triplanar corrections also are being used. For all of the various deviations of the foot, there are specific types of corrections for each type of deviation. If the deviations are not taken into consideration at the time of casting, the mold will not have the necessary corrections.

Mold Corrections and Cast Modifications

Once a patient’s limb is molded, it is corrected in the laboratory. The practitioner must carefully consider the areas to be corrected on each mold by evaluating each patient’s deformity. CMT is asymmetrical, so the left leg can have one deformity and the right leg another. So, when correcting CMT deformities, each leg should be treated separately with its mold modified and corrected to that limb’s deformities.

PRESCRIPTION CRITERIA

Often, physicians do not provide enough description in their prescriptions for AFOs. Physicians commonly will state “AFO” and designate the right or left limb with no indication regarding the need for a custom model or one from off the shelf. When writing a prescription for an AFO, the physician should consider the following: What should the orthosis accomplish? Does the patient need help with balance or a correction of a varus or valgus or other deformity? Does the patient exhibit a particular gait, get fatigued, have pain? Are they at a high risk for falls? Orthotists often will need to consult with the physician to make a recommendation based on what they observe. Ultimately, more standardized types of prescription criteria would be beneficial for physicians who need to prescribe orthoses, as they would be better able to indicate the orthotic goal and what needs to be accomplished with each brace.

TEST BRACING

Employing the method of test bracing, an orthotist will create a temporary permanent-style brace to use as a diagnostic brace. The test brace features the geometry and modifications that will appear in the definitive brace, but it is constructed of less expensive materials that do not have the same stiffness as those in the definitive brace. As the name implies, test bracing is intended to test the patient’s fit, comfort, balance, corrections, stability, and, to some extent, walking. Test bracing generally is undertaken for patients who need an energy-storing brace. When an energy-storing brace is completed, its structure cannot be changed. Therefore, the orthotist will want to ensure that all of the requirements are addressed prior to constructing the final brace. The fabrication of diagnostic devices prior to fabrication of an artificial limb is standard practice and is covered through health insurance. Unfortunately, this coverage does not exist for the orthotic (CMT) patient.

BRACING TECHNOLOGY OF THE FUTURE

There are many improvements that can be incorporated into the design of leg braces that are mechanically possible. This author has found that energy-storing technology is quite valuable to patients.
Many patients state that it reduces fatigue levels and that they can walk faster without getting tired. In the future, microprocessors may allow the ankle joint to move more like an anatomical ankle joint in that its computerized technology makes the decisions for that joint to move, much like a bionic knee. However, at this time, due to the size, complexity, and cost of such an improvement, there is no current technology to meet these demands.

**ENERGY COST OF WALKING**

CMT patients have a much higher energy cost of walking than the average individual. Patients complain of fatigue, abnormal walking, and decreased speed. Many clinical practices observe that these patients get tired at the end of the day. Also, the patients are overexerting healthier muscle groups due to gait compensations. These muscles include the hip flexors, as discussed earlier, and the back muscles to hip hike and flex the lower limb in order to clear the ground. CMT1A patients show a decline in motor performance due to loss of muscle strength and experience fatigue, foot and ankle deformities, alterations of balance, reduction of functional aerobic capacity, and, as a consequence, lower levels of daily activity.⁸

**EARLY FATIGUE**

CMT patients fatigue much faster than healthy individuals. Well-documented studies—having taking into account different walking speeds, the length of each individual’s step, and cost of energy—show that patients impaired with CMT do walk slower and at a higher cost of energy. In a recent study of CMT1A patients, it was stated: “In conclusion, the homogeneous group of CMT1A patients with low level of walking impairment selected for this study showed a greater metabolic and cardiac cost of walking per unit of distance when compared with healthy individuals.”⁴

There are some AFOs that are designed to reduce fatigue, such as carbon fiber energy-storing devices (e.g., Helios® dynamic release). They encourage a faster gait, create a faster swing phase, and a more normalized gait by allowing the patient to get their heel on the ground quicker, thereby reducing gait compensations.

**PATIENT EXPECTATIONS**

What a patient gets a new brace or a high tech brace, their expectations may exceed what is possible. Although a cure for CMT does not exist, most CMT patients are hopeful that an AFO will solve all of their lower limb deficiencies. Unfortunately, as we know, a brace has no motor. The patient is the motor and if the patient has muscles that are severely affected, or they are heading towards the fulltime use of a walker or wheelchair, that brace will not help them as much as the patient who does not need a walker or a wheelchair.

**GOALS FOR SUCCESSFUL BRACING**

The goal for a successful CMT brace is that it should give back as much function as possible to the patient. With that, it should restore as much balance as possible and it should correct as much deformity as possible. When a brace successfully corrects and stabilizes, it is then that positive functional outcomes occur. Out of all these considerations, the author’s experience indicates that balance is of the utmost priority to the average CMT patient. When evaluating patients, or even in initial consultations, the patient always requests help to stand without holding onto anything and then they would be very happy with their AFO. Most of these requests come from patients who already have AFOs and are not receiving that benefit. Why is this? Unfortunately, there is no requirement in the healthcare field that orthopedic bracing corrects for balance control. It is more acceptable that a patient use a cane or a walker for balance loss. However, most patients do not want to use a walker; they want to be as independent as possible and they want to look normal in society. When an orthotist fashions a brace to correct for deformities, they also are attempting to get as much balance back as possible for the patient. If a patient cannot stand balanced, they cannot walk balanced. All of the above combined criteria should be incorporated into a lower limb brace to provide a functional and stabilized gait.

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INTRODUCTION

In 1886, the first discussions of Charcot-Marie-Tooth (CMT) were introduced by Jean-Martin Charcot, Pierre Marie, and Howard Henry Tooth. Their studies opened the door to a better understanding of this disease. Since then, CMT has been studied as a progressive genetic peripheral nerve disorder that results in the degeneration of a neuron’s myelin sheath and axon. Normally, the myelin sheath allows rapid and efficient transmission of impulses along the nerve cells. The axon is a long slender projection of a nerve cell, or neuron, that conducts electrical impulses away from the neuron’s cell body (Fig. 1). When the myelin sheath and axon are disrupted by CMT, the frequency of signal transduction and impulses slow down, causing muscle atrophy from poor nerve signal transmission. CMT has been labeled one of the most common inherited neurological disorders and also can be identified as hereditary motor and sensory neuropathy (HMSN), which is defined as a group of disorders causing alterations in peripheral nerves. The different forms of CMT have various impacts with longterm and further disabilities, but recognizing the signs and symptoms early, along with implementing an efficient mode of treatment options, will allow the patient to live a much more comfortable, sustainable lifestyle.

DISEASE SYMPTOMOLOGY AND DIAGNOSIS

CMT currently affects around 125,000 people (mostly males) in the United States. There is no specific race that is susceptible to this disorder. Patients begin experiencing signs and symptoms around the age of 30, including clumsiness, foot pain due to their shoes, frequent falls and ankle sprains, as well as walking or gait abnormalities. For the patients who report these symptoms, claw toes, hammertoes, pes cavus, equinovarus, calcaneovarus, and foot drop are observed under clinical examination. Common appearances of the patient’s legs include an “inverted champagne bottle” and “stork’s legs” (Fig. 2). The most prevalent feature of CMT is a cavovarus deformity, which is a type of clubfoot characterized by an overly high longitudinal arch and an inwardly turned heel.

There are various ways of diagnosing CMT. Deep tendon reflexes test the ability of the nerve to stimulate muscles to produce a reflex
CHARCOT-MARIE-TOOTH DISEASE: CURRENT TOPICS AND TREATMENT OPTIONS

affect. When these are decreased, further tests can be administered to further support the diagnosis. Nerve conduction velocity, resistance strength tests, nerve biopsies, and genetic testing are among the most common modalities of diagnosing CMT.

Several stages of CMT exist including CMT1, CMT2, CMT3, CMT4, and CMTX. CMT1 is caused by abnormalities with the myelin sheath and includes 50% of CMT cases. CMT1 is further broken down into three subdivisions: CMT1A, CMT1B, and CMT1C. Roughly 80% of those fall under CMT1A, which is an autosomal dominant form of the disease in which there is a duplication of the gene located on chromosome 17 that directs production of a protein called myelin protein 22 (PMP-22). This is a vital portion of the myelin sheath. An overabundance of the gene leads to an abnormal function and structure of the sheath. The end result is atrophy and weakness of muscles in the lower extremity that began in adolescence.

CMT1B accounts for 5-10% of CMT cases. It is an autosomal dominant form of CMT in which there is a mutation (mostly point mutation) of the gene that directs the manufacturing of myelin protein zero (P0). Patients with CMT1B experience symptoms similar to those who have CMT1A.

The gene for CMT1C has not yet been identified, but the symptoms are also the same as those of patients with CMT1A.

Twenty percent of CMT cases fall under CMT2, which is caused by abnormalities of the axonal portion of the neuron. CMT2 is further broken down into subgroups ranging from A-L and factors that delineate them depend on mode of inheritance and clinical features. There is current ongoing research to better understand CMT2.

Dejerine-Sottas disease falls under the category of CMT3, which is a rare and severe demyelinating neuropathy. There are point mutations on P0 or PMP-22 that begin in infancy, resulting in the atrophy of muscles, weakness, and sensory problems.

CMT4 is an autosomal recessive form of CMT. The primary gene responsible has not yet been identified, but this form of the disease causes leg weakness in childhood. The patient later becomes nonambulatory once they reach adolescence.

CMTX covers 10-20% of all cases. It is caused by a point mutation in the connexin-32 gene on the X chromosome. Males who inherit one mutated gene from their mothers show moderate-to-severe symptoms of the disease beginning in late childhood or adolescence (the Y chromosome that males inherit from their fathers does not have the connexin-32 gene). Females who inherit one mutated gene from one parent and one normal gene from the other parent may develop mild symptoms in adolescence or later, or they may not develop symptoms of the disease at all.

WORKUP AND SURGICAL TREATMENT OPTIONS FOR THE VERY YOUNG TO ADULTS

When the decision is made to treat a patient CMT surgically, it is important to factor in several elements including, which of the remaining motor units are functional, how flexible or rigid are the established deformities, and is there ligamentous laxity present.

All deformities present in patient’s feet due to CMT must be corrected. Some of these include a combination of the following:

- Metatarsal deformities
- Midfoot deformities
- Forefoot deformities
- Rearfoot deformities
- Any ankle/equinus deformities
- Foot drop deformities
- Toe/digit deformities
- Tendon deformities
- Muscle imbalances

Correcting these deformities before they progress further will have a significant positive impact on the surgical outcome.

The National Institute of Neurological Disorders and Strokes is one of many research institutions currently striving to identify mutant genes and proteins that lead to the various CMT subtypes. Androgen has shown some progress to slow nerve degeneration in patients.

Many patients who have a flexible form of a cavovarus deformity (Figs. 3 and 4) and plantar flexed first ray (Fig. 5) associated with CMT have been surgically treated with reconstruction. The long-term results from a study have shown positive outcomes for those who underwent reconstruction with having a lower progression of degenerative changes in comparison of those who had a triple arthrodesis (multiple fusions instead of reconstruction). The goal of the reconstruction compared to the arthrodesis was to efficiently correct the cavus deformity with regard to function, radiographic changes, and gait/walk patterns. A triple arthrodesis procedure was concluded to be left for a last resort procedure when all others would be deemed to fail. The triple arthrodesis procedure has greater chance of alignment errors along with various forms of immobility issues.
When initially diagnosed, CMT patients generally are told that there is no cure for their disease. However, there are alternatives available to minimize the manifestations of the disease. Physicians have been able to treat CMT patients symptomatically, through physical and occupational therapy as well as by managing pain, fatigue, and orthopedic issues.

CMT patients can benefit from physical therapy in various ways, especially when working towards relieving functional deterioration. Not only does strength training help improve function of weakened muscles but it also helps to maximize the strength of uninvolved muscles. Studies show that even a small increase of strength in affected muscle can result in significant improvement for patients. This leads to greater tolerance of activities such as writing and ambulation. It is important to note that the results from physical therapy will differ from patient to patient. Progress can be slow for some, while for others it may produce a major increase in muscle strength and resistance. Unsure of the ultimate outcome, CMT patients may find physical therapy an exhausting process; however, compliance is essential for significant improvements.

Many patients with CMT become sedentary, which causes not only deterioration of the muscles but also deterioration of their overall health. Although, excessive training is contraindicated in CMT patients due to potential injury during training, studies show that mild-to-moderate training can improve overall strength and health by reducing heart disease and body fat as well as lowering blood pressure all the while increasing muscular and cardiovascular endurance. It is important to consult a physical therapist who can design an exercise program for an individual’s specific needs and thus help to avoid injuries. It is also important to manage fatigue in CMT patients because they expend more energy during exercise than that of a comparable healthy individual. The early stages of physical therapy can have a dramatic effect in delaying nerve deterioration and muscle weakness.

Utilizing orthotic devices, such as ankle-foot orthoses, during physical therapy to support the weakened joints and muscles also is an important part of improving a patient’s life. Dorsiflexors in the lower extremity are often weakened in CMT patients. As a result patients, may experience foot drop and can trip and fall easily. Proper orthotic devices can greatly reduce the chance of tripping and will help reduce injuries during physical therapy.

Some CMT patients experience weakness in their arms and hands, causing difficulty with gripping and finger movement. Occupational therapy can make a significant difference in the quality of life by using assistive devices such as special rubber grips on doorknobs or clothing with snaps instead of buttons.

CMT is a lifelong disease which requires support of various individuals throughout the course of the disease. Although it is not easy, proper treatment can significantly improve a patient’s quality of life.

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REFERENCES

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Charcot-Marie-Tooth
CME Questions

1. Which of the following deformities of the foot are associated with many of the hereditary motor sensory neuropathies?
   A. Cavus deformity.
   B. Equinus deformity.
   C. Tirus planus deformity.
   D. A. and B.

2. In Charcot-Marie-Tooth (CMT)1a, scoliosis can occur in up to __% of patients:
   A. 30%.
   B. 50%.
   C. 80%.
   D. 100%.

3. The Coleman block test is used to evaluate:
   A. Contracture causing equinus deformity of the ankle.
   B. Rigidly of hindfoot varus in a cavus foot.
   C. Stiffness of scoliosis.
   D. Brightness of a Coleman® lantern.

4. The preferred method of treatment of a flexible cavovarus deformity of the foot in CMT is:
   A. Triple arthrodesis.
   B. Bracing.
   C. Tendon transfer and osteotomy.
   D. Amputation.

5. The best position for the foot in CMT is:
   A. Plantigrade.
   B. Equinus.
   C. Cavus.
   D. Calcaneus.

6. The most prevalent form of CMT disease, CMT1a, accounts for approximately what percentage of all cases of CMT?
   A. 30%.
   B. 50%.
   C. 70%.
   D. 90%.

7. What is the minimum duration of resistance exercise training the literature identifies in terms of reliable effectiveness in the management of CMT disease?
   A. 6 weeks.
   B. 12 weeks.
   C. 18 weeks.
   D. 24 weeks.

8. What does the literature identify as the most important potential effect of resistance training in those CMT patients capable of participating in such an exercise program?
   A. Improved performance of activities of daily living.
   B. Hypertrophy of muscle fibers.
   C. Improvement of aerobic capacity.
   D. Improvement of the gait.

9. The literature indicates that CMT patients most likely to benefit from resistance exercise possess a minimum of what percentage of normal maximal isometric strength prior to participation in a training program?
   A. 15%.
   B. 30%.
   C. 50%.
   D. 75%.

10. The clinical goal of the Exercise Is Medicine® model is to:
    A. Encourage all allied health practitioners to participate in regular exercise.
    B. Adapt the exercise programs of general, otherwise healthy populations to persons with chronic disease, such as CMT.
    C. Create alternative treatment options in the medical community that can be reimbursed by third-party providers.
    D. Incorporate activity assessment and appropriate exercise prescription as standard clinical operating procedure in the prevention and treatment of chronic disease.

11. Bracing for CMT should address corrections for:
    A. Footdrop.
    B. Gastrocnemius weakness.
    C. Loss of balance.
    D. All of the above.
12. CMT patients who demonstrate the gait deviation of hip hiking are doing that due to the muscle weakness of the:  
A. Quadriceps.  
B. Hamstrings.  
C. Tibialis anterior.  
D. Tibialis posterior.

13. Corrective molds should incorporate:  
A. Shoe size.  
B. Physician’s prescription.  
C. Realignment of joint deviation.  
D. Measurement of pelvis width.

14. Which gait deviations cause the CMT patient to produce a steppage gait?  
A. Circumduction.  
B. Hyperextend gait.  
C. Hip hiking.  
D. Extensive knee flexion.

15. The most common deformity observed with CMT patients is:  
A. Pes cavus.  
B. Valgus.  
C. Equinovarus  
D. Pes planus.