Podiatrists' Role in Charcot-Marie-Tooth Disease

Many patients with CMT are never diagnosed.

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Introduction

Charcot-Marie-Tooth disease (CMT) is the most commonly inherited neurological disease, with an incidence equal to that of multiple sclerosis (1 in 2,500 people). There are currently 2.8 million people with CMT—so why don't podiatrists treat more people with this disease? With over 70 genetic variations of CMT now identified, you might wonder how, as a busy clinician, you could ever initially diagnose CMT without specialized blood work and consultation with a neurologist. Oftentimes, the subtle signs are there in a "routine" visit for a painful callus, sprained ankle, hammertoes, or falling and/or being clumsy. The progressive nature of the disorder complicates planning and treatment.

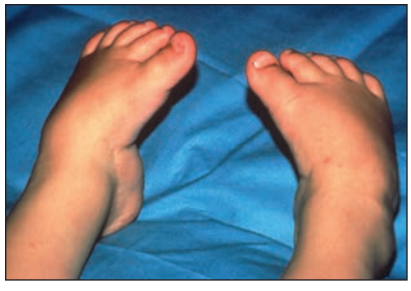

Figure 1: Early flexible deformity with muscle imbalance.

Presenting Complaints

Children may be brought in by their parents with the complaint of the child being clumsy, not good in sports, or having a funny-shaped foot. Most CMT-afflicted individuals present with a cavus foot of varying degrees (there are subtypes with a flat foot, but those are beyond the scope of this article). Fungal toenails are quite common in the CMT population, so this may be the portal by which your patient finally gets a definitive diagnosis of "funny feet" (Figure 1).

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Hammertoes are common complaints of those with CMT, as well as ulcers (typically neuropathic) in various plantar and digital high pressure locations, depending on deformity and progression of weakness and muscle imbalances. Claw toe type hyperkeratoses are common (Figure 2), and can be painful despite having peripheral neuropathy. Neuropathic pain similar to what one finds in diabetic neuropathy is common; management is similar to those with diabetes pain, and often requires a team approach with a physiatrist or other pain management specialist.

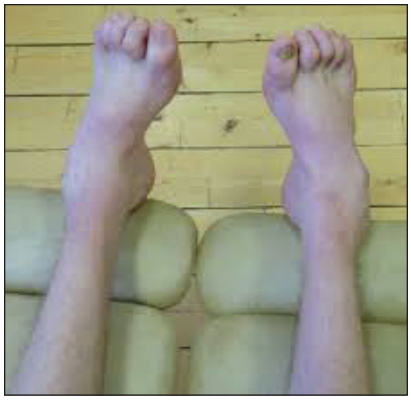

Figure 2: Adducto-varus with claw toes, prominent lateral talar head.

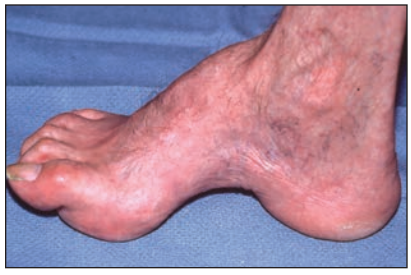

Figure 3: On weight-bearing there is minimal change in arch height or length.

Clinical Exam

A vascular exam may be normal; oftentimes, pedal pulses are intact and symmetrical. Usually, one will see signs of lymphedema or venous congestion, frequently worsened by prolonged wearing of braces and/or shoes with minimal foot elevation and activity (Figure 3).

A key diagnostic marker is the hallux hammertoe; commonly, this is a key to differentiate between a neurologic cavus foot and just a "high arch." As the intrinsic musculature of the feet progresses, the short flexor to the hallux weakens and the long flexor pulls

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the distal phalanx plantarly, leaving a prominent hallux I-P joint. Test the forefoot intrinsic muscles by asking the patient to fan, or spread the toes open. The examiner may also interlace his/her fingers between the patient's toes and assess squeeze strength.

Early on in the progression, a little heel varus is noted (Figure 4), with flexible hammertoes; but weakness may have already started in the anterior tibial and extensor longus muscles, giving you a further diagnostic differential, an easy way of assessing heel walking strength (loss of full dorsiflexion power, i.e., dropfoot), and you can begin to focus on a provisional diagnosis of CMT. Many times, there is no documented family history of CMT or neurologic disease per se; typically, relatives may have had a limp, or used a cane for some previously unknown reason. Look for the classic "inverted champagne bottle" calf, as well as underdeveloped muscles in forearms, and a flattened thenar eminence in the palmar hand (Figure 5).

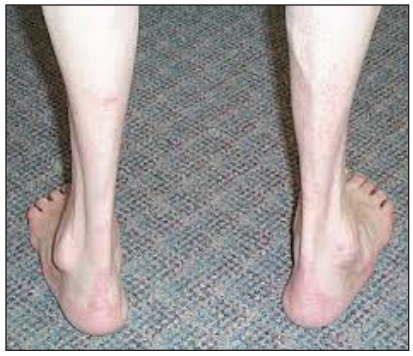

Figure 4: Cavo-varus with prominent lateral malleolus.

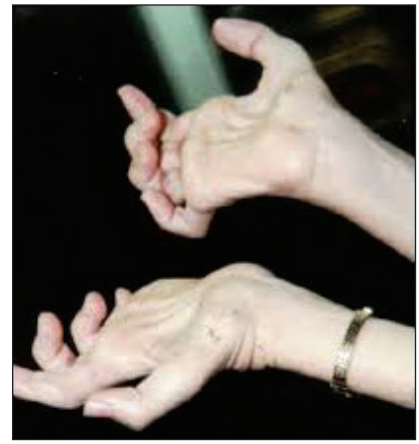

Figure 5: Intrinsic muscle loss with destabilization in palmar hand.

Neurologic exams typically reveal minimal to no DTRs in the upper and lower extremities. While sharp/dull and Semmes-Weinstein testing can be normal until late stages in the disease, vibratory sensation is oftentimes impaired fairly early. The epidermal nerve fiber density (ENFD) biopsy can be helpful as an initial screening device when attempting to make the provisional diagnosis of CMT.

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Radiologic Exam

Standing A-P and lateral views will likely reveal an increased calcaneal pitch, plantarflexed 1st ray, and forefoot equinus. Metatarsus adductus is common, along with hammertoes of varying degrees. Many times, on the lateral view, one can appreciate the ankle mortise resting in a near fully dorsiflexed position.

Etiology

Distal muscle wasting secondary to nerve pathology brings about a unique clinical presentation. The interossei, lumbricales, and other intrinsic muscles such as the flexor hallucis brevis and extensor hallucis brevis are weakened; this leaves the lesser affected longus muscles (with origins more proximal, in the calf) to overpower the intrinsic muscles' stabilizing forces normally present.

As the disease progresses, typically the lateral calf muscles (evertors and extensors) become affected first. This leads to the inverted and varus nature of the boney structure in the feet. Varying degrees of foot drop may be present, as well as decreased peroneus brevis strength. Lack of opposition to the peroneus longus will cause an increase in plantarflexion of the 1st ray, as the extensors and anterior tibial muscles weaken.

Overall, a shortening of foot length occurs as a "plantar folding" of the midfoot occurs, with an adducto-varus component. Increased calcaneal pitch follows, with forefoot equinus masquerading as Achilles tendon contracture. This is a good reason NOT to perform tendo-Achilles lengthenings on CMT patients.

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Laboratory Tests

After performing an ENFD exam, most podiatrists will refer to a neurologist or physiatrist for nerve conduction velocity (NCV) and EMG tests. It is not unusual to see distal latencies in the single digits with demyelinating types of CMT, with a profound slowing of the nerve signal. Axonal pathology reveals a dampening in the peak nerve pulse height. Hopefully, you have a relationship with a neurologist or physiatrist who is skilled in these exams without causing significant pain to your patient. Sural nerve biopsies are rarely done today.

Once the diagnosis is confirmed, it is valuable to consult with your patient on the blood tests available for the actual genetic phenotype of CMT the patient has. Several blood labs now offer the

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DNA analysis necessary to determine which chromosome has the mutation, and therefore help in family planning, as well as typical clinical course with that type of CMT. Athena diagnostics will do reflexive DNA testing, starting with PMP22 duplication/deletion testing for the most common cause of CMT.

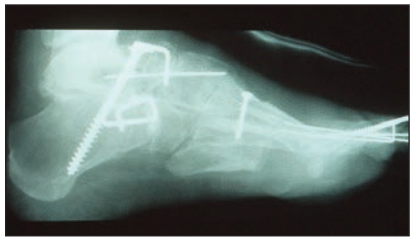

Figure 6: Triple arthrodesis with forefoot wedging, digital fusions, partial 5th metatarsal shaft excision-decrease forefoot pressure, and help with hindfoot alignment.

Conservative Treatment

These patients need supportive shoes, usually high top or, if you also prescribe an AFO, an added depth low cut shoe. Palliation is paramount, and accommodative insoles seem to work best to prevent pressure build-up. There are a few good custom accommodative orthoses made today. What seems to the podiatrist like a miniscule amount of plantar hyperkeratosis can cause incredible pain with ambulation; a quick debridement is heaven for these people.

With the advent of OTC carbon graphite dropfoot braces, many times podiatrists can be heroes without a lot of headache. Custom AFOs may be required for significant deformity or profound differences in girth size between calf and ankle or foot. Rocker bars/metatarsal bars work well for early drop foot, as do shoe modifications like a lateral Dutchman—which can help stabilize over-supination.

Newer technologies have helped create variable density wicking socks which not only cushion, but help with the sudomotor changes typical with neuropathy. Toe spreaders, like Correct Toes or Yoga Toes, can help with the oftentimes cramped, sometimes painful, but mostly paralyzed lesser toes. Foot strapping and taping are helpful, whether the classic low-Dye taping or newer techniques with waterproof stretch tape that can help with alignment and comfort.

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Prescription medications like Lyrica and gabapentin play a role in controlling pain, when present (not all CMT patients have pain, however). The nutritional supplements popular for diabetic neuropathy (benfotiamine, alpha lipoic acid, B12) can help CMT neuropathy as well.

An exercise program emphasizing ROM, stretching, and gentle strengthening is highly recommended; Pilates, Gyrotonics, Yoga, and some types of Tai Chi are particularly helpful for balance disorders like Charcot Marie Tooth. Newer technologies like the Neurogenx and TENS units show promise to help with neuropathic pain.

Surgical Approaches

The most common complaints are painful claw toes and deformity causing plantar forefoot hyperkeratoses. Arthrodesis of involved joints (digital PIP and DIP joints) has been the gold standard (Figure 6); minimally invasive/percutaneous surgery with fluoroscopy may be a good option for smaller bone decompression. A Jones procedure with tethering of the extensor hallucis longus into the dorsal 1st metatarsal can be helpful for not only the drop foot, but for preventing the hallux from suffering pressure on a shoe dorsally, secondary to extensor substitution (Figure 7).

Cavus reconstructions are difficult to make look pretty and function well. Three-dimensional sliding osteotomies of the heel can be helpful for the heel varus and subsequent lateral ankle challenges seen in the untreated varus heel. Midfoot osteotomies are an option, but individual metatarsal dorsiflexion osteotomies can achieve many of the goals you and your patient desire, with typically faster healing and greater patient satisfaction.

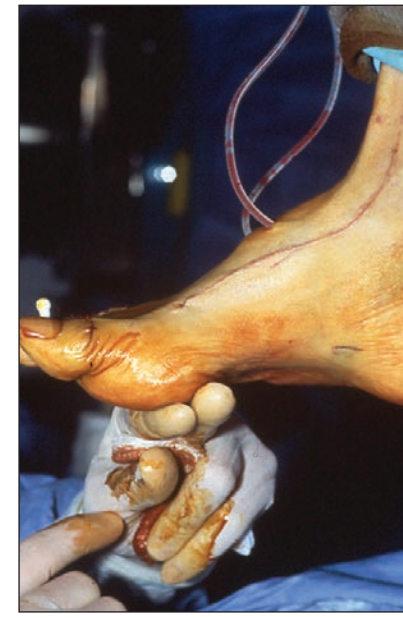

Figure 7: Jones tenosuspension and dorsiflexory osteotomy of 1st metatarsal.

Anastomosis of the peroneus longus (either en toto, or partial) to the peroneus brevis will oftentimes give greater lateral ankle stability, as well as removing some of the deforming plantarflexory force on the 1st metatarsal head that most cavus sufferers have. Steindler stripping can be quite effective, although inadvertent soft tissue damage is common.

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The posterior tibial muscle oftentimes remains strong until late in the disease (Figure 8). The surgeon should consider lengthening the muscle, or transferring it dorsally to help with foot drop.

Support Organizations

The Charcot Marie Tooth Association (CMTA) is probably the largest and most influential of the organizations (disclaimer: this author is a member of the CMTA Advisory Board). They maintain local support group meetings in most U.S. states. Volunteering to teach to these groups can be very rewarding for the podiatrist. They direct funds toward research, and hopefully a cure. HNFCure.org also directs research funds for hereditary neuropathy and has Team CMT, a sports-based fund raiser group. Both have newsletters via email, and they want more input from the podiatric community. CMTA has also established CMT Centers of Excellence in the U.S. and three other countries. Drs. Michael Shy and Steven Scherer have assembled teams with great knowledge of CMT, but as of yet, not a single podiatrist is involved in these Centers of Excellence, with the exception of the Australian Center.

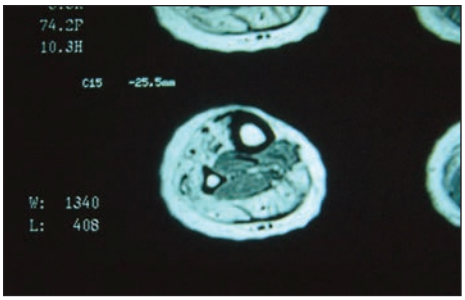

Figure 8: Transverse image of left calf. The deep flexor muscles spared of atrophy.

against the various types of CMT in the hopes of then beginning FDA-approved drug trials soon.

This all needs money, and a podiatric consortium to solicit donations would be great for our profession as well as helping with drug therapy for CMT. The Hereditary Neuropathy Foundation has partnered with another philanthropic organization (BioPontis Alliance) to also generate research into drug therapy models.

Podiatrists need to play a key role in Charcot-Marie-Tooth disease diagnosis and management.

Research

There are at least 70 different chromosomal abnormalities which cause CMT. These obviously all have slightly different phenotypes, but all are grouped under the umbrella of CMT. Type 1 is still the most common type and typically exhibits the cavus foot type we have all seen before. The STAR initiative (Strategy to Accelerate Research) has been driven by CMTA and works with the NIH and several prominent neurologist researchers around the country. Via the use of high-throughput labs, millions of drug compounds can be tried

Summary

Podiatrists need to play a key role in Charcot-Marie-Tooth disease diagnosis and management. With the advent of newer diagnostic biopsies and blood work, we will increase our value as a profession by becoming even more integrated team players with the neurologists, physiatrists, orthotists, PTs, and orthopedic surgeons to best manage these challenging lower and upper extremity manifestations. **PM**

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Dr. Stilwell retired from surgical podiatry practice in 2013 secondary to effects of CMT on his hands. Now as president of Hozhoni Health Services, LLC he is beginning to manufacture and market his patented Barefoot Orthotic™ to podiatrists' offices exclusively. Hozhoni is a Navajo term for walking in balance; gait analysis and podiatric therapies for those with CMT is Hozhoni Health's mission statement.

Dr. Dan Stilwell is a licensed Podiatrist interested in the fusion of Alternative and Western medicine in the treatment of common foot and ankle pathology. His intention is to address each problem from a Holistic standpoint through patient education and preventative medicine. He uses a variety of treatments such as laser therapy for toenail fungus, homeopathic injections, pole-walking for gait training and assistance, and the use of the patented Barefoot Orthotic. Dan received his Bachelor of Arts in Spanish with a Minor Emphasis in Chemistry from Northern Arizona University in 2008. He graduated from the California School of Podiatric Medicine in 2012, and in 2015 completed a 3-year Podiatric Surgery and Medicine Residency at Intermountain Medical Center in Salt Lake City, UT.