Practice Brief

Synthesising research evidence to inform practice

Neuro-muscular Disorders

NUMBER 1. Allied Health & Nursing Alliance series

July 2015

Charcot-Marie-Tooth disease: why feet matter

With the recent explosion in gene discovery, many more people are being diagnosed with Charcot-Marie-Tooth disease (CMT) and foot care is at the forefront of their management. While allied health professionals such as podiatrists, physiotherapists, orthotists and pedorthists have the skills to treat those with CMT, they may lack the necessary knowledge or confidence to treat the condition. This **Practice Brief** aims to help by providing an outline of the disease and the most effective ways to treat the feet of someone with CMT.

What is Charcot-Marie-Tooth disease (CMT)?

Charcot-Marie-Tooth disease (CMT) is the collective term for a group of clinically and genetically diverse nerve disorders. CMT is the most common inherited peripheral neuropathy and affects around 1 in 2,500 people.[1] The disorder is named after the three neurologists who described it in the late 1800s. CMT is also known as **Hereditary Motor and Sensory Neuropathy** (**HMSN**) and can be caused by more than 80 different genetic variations.

CMT is most commonly characterised by a painful, high-arched foot deformity (pes cavus) (Figure 1).[2] However many people with CMT will also experience distal weakness causing foot drop, sensory loss, absent tendon reflexes (especially ankle), muscle cramps and other foot deformities such as hammer toes (see Table 1 for reported frequency of CMT signs & symptoms).[2-4]

As there are multiple forms of CMT, the onset and severity of symptoms varies. Some individuals develop severe disability in infancy or early childhood (e.g. Dejerine-Sottas neuropathy; CMT1E), whereas others develop few if any symptoms of neuropathy until adolescence or adulthood. The most common form – CMT1A, progresses slowly but other forms present earlier, progress faster and limit life span.

CMT is a lifelong condition which can affect both the occupational and social-emotional aspects of a person's life. The symptoms most likely to affect a person's quality of life are weakness, muscle cramps and pes cavus.

Figure 1. Pes cavus in a child with CMT

Why is this issue important?

Allied health professionals are often the first clinicians to 'spot' CMT and can facilitate early diagnosis.

Allied health professionals are often the first clinician a child (and some adults) with undiagnosed CMT will see. This is because the first symptoms of CMT are often high-arched feet (pes cavus), lower limb weakness (resulting in trips and falls), and difficulty running and keeping up with peers. This means that allied health professionals are uniquely positioned to ensure patients get a confirmed diagnosis early on from a neurologist.

Allied health professionals are an important part of the team caring for someone with CMT.

Allied health professionals will most likely see someone with CMT regularly, as CMT is a life-long condition that mostly affects the extremities of the limbs. Collectively allied health professionals provide important primary care (e.g. maintaining good toenail and skin health), physical therapy (e.g. stretching and strengthening exercises), devices/appliances (e.g. foot and ankle orthoses, footwear provision, advice, and adjustment) and education (e.g. hosiery and care procedures to do at home).

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Allied health professionals have the skills to treat individuals with CMT but may lack confidence or the knowledge to do so.

Podiatrists, physiotherapists, orthotists and pedorthists often have little formal education or training in caring specifically for those with CMT. As a rare disease, allied health professionals may also not have seen many patients with CMT. This can lead some practitioners to feel unsure about how to best care for the CMT patient.

Assessing people with CMT

At first, the peripheral neuropathy causes selective weakness in the feet and lower legs.

Typically this involves lateral ankle instability (e.g. ankle sprains and tripping), and foot posture changes towards pes cavus. Planovalgus flatfoot deformity can also occur in cases of generalised lower leg weakness and ligamentous hypermobility. It's these foot and ankle symptoms that first lead most people with undiagnosed CMT to seek health care and become aware of their disorder.

As the weakness associated with CMT progresses, a characteristic foot drop gait develops.

This develops because the person's anterior lower leg muscles cause the foot/ankle to drop during the swing phase of gait and makes it hard for them to clear the ground with each step (it can also develop into a 'steppage-gait' or high stepping gait due to increased hip and knee flexion).

You can investigate muscle weakness initially by asking the person to walk on their heels. The weakness of the anterior and lateral muscle groups (and associated ankle contracture) makes this almost impossible for those with CMT. A child with CMT will also have progressive difficulty with sports, and may experience weakness, pain and tremor in the hands.

It's a good idea to refer a patient you suspect of having CMT to a neurologist if:

- they have a high foot arch (pes cavus), and high arches run in their family; and
- they have weak ankle dorsiflexion strength and/or difficulty walking on their heels
- their ankle-tendon reflexes are absent

Table 1. Reported frequency of signs and symptoms among children and adults with CMT. [2-4]

Signs and symptoms	Frequency in 81 children with CMT1A aged 2-16 years [2]	Frequency in 528 children with all types of CMT aged 3-20 years [3]	Self-reported frequency in 295 adults with all types of CMT [4]
Weakness of legs/feet	100% foot dorsiflexion weakness	93% foot dorsiflexion weakness	80%
Reduced sensation	-	Pinprick: 35% Vibration: 34%	93%
Pes cavus	11% (2-6 years) 63% (12-16 years)	11% (3-7 years) 27% (8-12 years) 48% (13 years+)	80%
Poor balance	-	58%	95%
Difficulty heel walking	84%	85.9%	-
Ankle equinus	80%	-	-
Foot pain	27%	40%	-
Foot/leg cramps	36%	35%	79%
Ankle instability during walking	72%	53%	-
Frequent tripping/falls	63%	42%	-

Treating CMT

There is no cure for CMT. Current therapies focus on treating symptoms and can include podiatry, physiotherapy, occupational therapy and orthopaedic intervention. To date, there are no published comprehensive standards of care for CMT.

Successful prevention and treatment of pes cavus and associated foot pain and disability in people with CMT is also clinically challenging. While conservative and surgical approaches have been described in the literature, no firm evidence yet exists for any approach.[5] We do know however that foot and ankle orthoses, and stretching are beneficial.

Foot orthoses and specialised footwear are often effective in reducing and redistributing plantar pressure loading.[5]

The range of orthotic and footwear devices available is staggering, but this variety can also make it difficult to find a suitable combination of the two. It is important to discuss this challenge with each person, as well as the value of custom-made footwear and the types of shoes they prefer to wear. An evidenced-based prescription for an effective custom-made in-shoe device is shown in Table 2.[6]

Ankle-Foot-Orthoses (AFOs) can also assist depending on the level of deformity and disability.

Allied health professionals can provide guidance regarding the type of device for various stages of the disease. They can also provide information related to bracing for lateral ankle instability to prevent ankles sprains, trips and falls. A proposed clinical algorithm to assist the prescription of the correct foot and ankle orthoses for children with CMT is shown in Table 3.[7]

Stretching interventions are useful in improving ankle flexibility.

A major problem for people with CMT is the restricted ankle dorsiflexion range of motion, or ankle equinus.[2] Over time, the lower limb weakness that CMT causes leads to a shortening of the gastrocnemius and soleus, which can limit a person's activity, walking ability and postural stability. While prefabricated night splinting does not seem to be effective, serial night plaster casting has been shown to significantly increase ankle range in children and young adults with CMT.[8] There is little consensus in the literature for any other type of intervention to address ankle contracture.

Other rehabilitation approaches include strengthening of weak muscles, debridement of plantar callosities and strategies to improve balance.

Where a person has prominent sensory loss, expert primary foot care can also be particularly beneficial.

There are also numerous surgical procedures to correct and prevent the progression of the cavus deformity.

These surgeries rebalance and reconstruct the foot,[5] and can be divided into three main types:

1. soft-tissue procedures (e.g. plantar fascia release, Achilles tendon lengthening, tendon transfer)
2. osteotomies or removing a wedge-shaped portion of the bone (e.g. metatarsal, mid-foot or calcaneal);
3. bone-stabilising procedures (e.g. triple arthrodesis which fuses the three joints in the foot and ankle).

However, there is little evidence on the effectiveness of these procedures and few reports of long-term follow-up of patients who underwent foot surgery for CMT.

Several other whole body signs and symptoms can require careful clinical attention.

These include muscle cramps,[7] hip dysplasia and the complicating effect of co-morbidities such as diabetes.

Table 2. Comparison of the Prescription Guidelines and materials used for each intervention.[6]

Prescription	Custom Foot Orthoses	Control
Cast/scan modifications		Nil
Balance	Metatarsals 1–5	
Tissue expansion	20% medial arch expansion	
Corrected calcaneus position	0°	
Intrinsic forefoot	0°	
Shell shape		Nil
Length	Proximal to metatarsal heads	
Forefoot width	Standard (1–5)	
Lateral heel expansion	6 mm	
Heel cup height	12 mm	
Shell material	3-mm polypropylene	Nil
Shell posting		Nil
Extrinsic heel post	Lateral half only	
Motion	0°	
Elevation	0 mm	
Heel lift	0 mm	
Top cover		
Style	Full length	Full length
Material	3-mm Poron/ Kashmeer	3-mm latex foam/ Kashmeer

"Allied health professionals are often the first health professionals to 'spot' CMT and can facilitate early diagnosis."

Table 3. Clinical algorithm for the optimal prescription of foot and ankle orthoses for children with CMT.[7]

Impairments and activity limitations	Orthoses
Pes cavus and foot pain	Foot orthoses
Pes cavus and poor balance	UCBL* orthoses
Pes cavus and poorer balance (not corrected by UCBL* orthoses)	Supramalleolar orthoses
Pes cavus and poorer balance (not corrected by supramalleolar AFOs†)	Hinged AFOs†
Foot drop and poor walking	Posterior leaf spring AFOs†
Foot drop, poor walking, pes cavus, and poor balance	Hinged AFOs† with PF‡ stops
Global weakness of foot/ankle muscles and poor walking and/or balance (with/without pes cavus and/or foot drop)	Hemispiral AFOs†
Global weakness of foot/ankle muscles and poorer walking and/or balance (not corrected by hemispiral AFOs†, with/without pes cavus and/or foot drop)	Solid AFOs†
Pes cavus and/or ankle equinus (≥0°, not corrected by hinged AFOs† with/without PF‡ stops)	Solid AFOs†

Note: *University of California Biomechanics Laboratory; †ankle-foot orthoses; ‡plantarflexion.

What support is there for people with CMT and their treating health professionals?

There are a number of **CMT Centres of Excellence** (www.cmtausa.org) around the world. The Children's Hospital at Westmead in Sydney is currently the only specialty CMT clinic in Australia. The neuromuscular clinic at the Royal Children's Hospital in Melbourne also provides care for children with CMT.

The **CMT Association of Australia** (www.cmt.org.au) can provide information to individuals and their families on key health providers in each state, as well as metropolitan and rural points of contact.

Patient registries collect data that can be used for studies of CMT disease progression, to recruit patients for therapeutic trials, and to facilitate studies identifying genes linked to CMT. The international **Inherited Neuropathies Consortium** registry (www.rarediseasesnetwork.org/INC/register) is available to CMT patients in Australia.

If your patient is interested in joining a registry, you can contact the **Australasian Neuromuscular Network (ANN)** for further information (www.ann.org.au/contact-us).

Australasian Neuromuscular Network (ANN)

The ANN was founded in 2010 by members of the NHMRC **Centre of Research Excellence in Neuromuscular Disorders,** in partnership with other neuromuscular specialists, allied health professionals, scientists and patient advocates across Australia and New Zealand.

The ANN exists to enable those working in the field to share knowledge and work together to more effectively research, diagnose, treat and advocate for those with neuromuscular disorders.

www.ann.org.au

Things to remember:

1. Take a family history and refer a patient you suspect of having CMT to a neurologist.
2. Clearly discuss with your patient the goal of footwear or orthotic therapy (e.g. to alleviate pain, offload, increase walking ability, improve balance).
3. You have the skills to manage many of the symptoms a patient with CMT experiences, however consider how your patient might benefit from the care of other health professionals and where possible, connect with these professionals to devise an integrated patient-centred treatment plan.

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Conflict of interest disclosures: no conflicts of interest were reported.

Funding and support: Professor Joshua Burns is supported by grants from the NHMRC (National Health and Medical Research Council of Australia, Centre of Research Excellence #1031893, European Union Collaborative Research Scheme #1055131), NIH (National Institutes of Neurological Disorders and Stroke and Office of Rare Diseases, #U54NS065712), and donations from Muscular Dystrophy Association, CMT Association (USA), Australian Podiatry Education & Research Fund and CMT Association of Australia.

About us

The Centre of Research Excellence in Neuromuscular Disorders is a collaboration of neuromuscular experts.

It uses the latest approaches in medicine, science, nursing and allied health to improve diagnosis, facilitate prevention, and transform treatment from compassionate management to effective therapy.

The Centre is funded by the National Health and Medical Research Council.

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