FEATURE

THE REALITY OF CHARCOT-MARIE-TOOTH DISEASE:

HOW PODIATRISTS CAN HELP US

By Marilyn Kremmer

For this 'giving back' themed issue of STRIDE, we have partnered with the Charcot-Marie-Tooth Association Inc now known as CMT Australia, which supports people with this genetic disease.

As podiatrists are often the first health practitioners to treat affected people, CMT Australia wants to raise awareness amongst APodA members on how to best help a person with CMT. Here is Marilyn's advice to podiatrists, as told to STRIDE magazine.

> My wish is for as many podiatrists as possible to understand this condition. Podiatrists can make a big difference through their expertise, to make a person who has CMT feel 'understood'.

This is my story, which I share with members of the Australian Podiatry Association, to explain what it is like to have Charcot-Marie-Tooth disease, and how you can help.

I come from a 'CMT family' – my grandfather, mother, uncle, two sisters, our daughter and two nieces are all diagnosed. We all developed symptoms in our 40s. Until that point, we had active lives with no evidence of this disease.

However, I do remember watching my grandfather 'flop' around with a piece of tyre inner tube. He would cut it to make straps that linked from his shoelaces, and then he wound the straps around his ankle to keep his foot from flopping!

The experience of frustration

We have an unusual form of CMT – it is called Type 1B, with 'a few other things thrown in' as my neurologist tells me. It is the axonal variety – not the myelin variety.

While watching my mother develop foot problems, I saw the frustration it caused. She was finally diagnosed as having CMT by a specialist at the Royal Hobart Hospital.

At the time, we knew nothing about this disease. From her diagnosis onwards she tried to get help from the medical profession, who seemed to know very little about this disease. She would even attend doctor's exams as a 'guinea pig', to do her bit to raise awareness of CMT's symptoms amongst junior doctors.

The start of my symptoms

When I was in my late forties, my symptoms began to emerge. I started to notice the feeling of grit between my toes; it was like small pebbles in my shoes.

Despite having a family history of CMT, I was told that I had 'restless leg syndrome' by health professionals, and apparently, there wasn't much they could do about it. This was despite my twin sister and elder sister having already been diagnosed with CMT!

It wasn't until I requested a referral to a visiting specialist that I was finally diagnosed with CMT, following a nerve conduction test and later a genetic blood test identifying the type that I had.

Since I knew the frustration my mother experienced throughout her disease, I placed an invitation in our local newspaper to anyone with CMT to come to my house for afternoon tea. This was back in 2005. Five people initially responded to that first invitation. This has since grown to approximately 49 families that we know of in Tasmania. Support Groups are now in every state with 'Coffee and Chats' held regularly. CMT Australia also supports 'Aussie Kids' which runs camps for children with CMT and much more.

Our aim of CMT Australia today is 'to

From left to right: Marilyn with her sisters Diana and Julie, Christmas 2016. Sadly Julie, Marilyn's twin sister passed away following a stroke and complications from CMT.

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enhance the quality of life for people with CMT and those who care for them'.

Sharing patient experiences

At that inaugural 'coffee and chat' meeting it was clear just how frustrated we were when it came to the lack of knowledge in the medical community to help people with CMT.

Sadly, this frustration continues today. When seeking medical help, the response is usually along the lines of, "What is THAT?! Never heard of it." Yet it is the most common form of inherited neuropathy affecting approximately one in 2500 people. Not everyone who carries the gene becomes symptomatic, however.

The role of podiatrists

What I would love to emphasise to any podiatrists who may not be familiar with CMT is that while there are many forms of CMT, the common denominator is feet!

People who have CMT will inevitably need to visit a podiatrist at some stage for help. Podiatrists are often the first allied health professionals that people with CMT are referred to. It is likely to be a vulnerable time as the person comes to terms with their diagnosis, especially given CMT does not improve.

My wish is for as many podiatrists as possible to understand this condition. Podiatrists can make a big difference through their expertise, to make a person who has CMT feel 'understood'.

My podiatrist has made a real difference to my symptoms. For example, my balance became a real problem. Now, through the help of my podiatrist, I am able to stand and not 'wobble'. The podiatrist created custom made orthotics, which gave me a new confidence.

If only my mum had been referred to a podiatrist! Instead, she was made to wear ugly boots and callipers, 'leg irons' as we called them. Then it was up to the family to cut her toenails. She also loved having her feet massaged.

Steps you can take

If you are treating someone with CMT, or you have done so in the past, please direct them to the Charcot-Marie-Tooth Australia.

Here are some other ways that you or your patients can get involved:

- Attend the National CMT Seminar in September each year
- Head online to **cmt.org.au** (for details on the seminar, on CMT Australia and for a range of resources).
- Call the National Office on: **(02) 97675105**
- Email the team at: **cmtaa@cmt.org.au**

Marilyn with Tony Adams, President of CMT Australia along with two of our CMT Aussie Kids at the 2018 National Seminar held in Hobart.

FEATURE

Q+A with Professor Josh Burns

Can podiatrists diagnose CMT?

A neurologist diagnoses CMT, however, a podiatrist is often the first to notice early signs.

What can podiatrists look out for if they suspect CMT?

Look out for progressive pes cavus, absent tendon reflexes, sensory loss, abnormal gait such as foot drop, family history, inability to walk on heels, and an increase in trips and falls.

What is the 'best practice' podiatric treatment for CMT, or are there many options?

Podiatrists can look up the Practice Brief on the best foot orthoses for children and adults with CMT here: **mcri.edu.au/news/practice-brief-charcot-marie-tooth-disease.**

A Guide to Physical and Occupational Therapy for CMT may also be useful, located here: **cmt.org.au/uploads/pdf/CMTA_PT_OT_Guidel_9_2018_DT.pdf**

Where else can podiatrists go for more insights on CMT (apart from the CMT Australia website)?

All information relating to the assessment of children with CMT, aged three to 20 years, can be accessed here: **calculator.cmtpeds.org**. This includes related papers, manual of operations, YouTube videos, translations and so forth.

The 'Inherited Neuropathies Consortium' web page also provides information for health professionals, accessed here: **rarediseasesnetwork.org/INC**

Prof. Burns is the Associate Dean (Research) and Professor of Paediatric Neuromuscular Rehabilitation at the Faculty of Health Sciences, University of Sydney. He is also the Director of the Paediatric Gait Analysis Service of New South Wales which operates out of the Sydney Children's Hospitals Network (Westmead).

WHAT IS CMT?

CMT or Charcot-Marie-Tooth disease (named after the three medical professionals who first identified CMT), is also known as Hereditary Motor and Sensory Neuropathy (HMSN). It is a common but frequently undiagnosed condition. CMT is not a contagious disease but is an inherited neurological disease and in some families has a 50% chance of being passed onto other generations. Research has indicated that as many as one in 2,500 people in Australia could have CMT. CMT is not life-threatening, although people with CMT have it for life.

SYMPTOMS

CMT causes slow degeneration of the peripheral nerves including feet, legs, arms and hands. Typically muscles are weakened due to the loss of stimulation by the affected nerves. The severity of the disease can vary a great deal from person to person even within the same family. The symptoms are also varied. There are over 82 types of CMT with no known cure for CMT at this point in time. However, significant research is being done both in Australia and overseas to find one.

A common symptom of CMT is weakness in leg muscles which affects one's coordination and balance in such things as walking, running, walking up or down stairs, walking on uneven ground or just standing still for long periods. Other symptoms which may affect CMT patients include tremors, fatigue and diminishing fine motor skills.

Foot abnormalities such as high arched feet or, in some cases, flat feet, weak ankles and tendon tightening pose the most serious problems. Sometimes patients may need to wear foot orthoses or braces or undergo surgery. Even the simplest things, functions we take for granted, like unscrewing a bottle top, fastening buttons, turning on a tap or opening a door handle can pose problems for those with muscle wastage in the hands.

Untidy handwriting, due to not having control over the muscles and nerves in the hands and fingers, can pose a big problem for both children and adults.

CMT Australia strongly encourages people with CMT to participate in as many regular daily activities as possible and to develop skills in areas that they can excel in.

THE GOOD NEWS

Significant progress is being made regarding CMT management strategies that have the potential to provide a fulfilling and healthy life.

Thank you to CMT Australia and Prof. Josh Burns for giving permission to republish the contents of this brochure. ■