CMTAA
Charcot-Marie-Tooth Association Australia Inc.

I Have Been Diagnosed With Charcot-Marie-Tooth Disease What Happens Next?

Scenario

"I was very active and certainly not the fastest person when it came to running. Additionally, I was 'bumble footed' and could easily trip over as my walking gait was not a 'heel – toe' action.

As time went by, it was time to go to my GP who referred me to an Orthopaedic specialist who referred me to a Neurologist.

I was told that I had a neuromuscular condition known as Charcot –Marie-Tooth Disease (named after the 3 doctors who discovered this disease), also known as Hereditary Motor and Sensory Neuropathy (HMSN). I was advised that it was not life threatening, it is degenerative, and to get on with life!"

You may be able to relate to this or have a similar story.

The reality of this may be summarised by the following 3 important points:

1. **We must Take Responsibility For Our CMT**

2. **We Need To Have CMT Specific Knowledge**

3. **We Need To Manage Our CMT**

Let's look at these in more detail.

1. We must Take Responsibility For Our CMT

This is most significant. The reality of the situation is that if we don't take responsibility for our CMT it is most unlikely someone else will. We need to have our own determination and drive to follow through with this important responsibility.

2. We Need To Have CMT Specific Knowledge

We want to know who and where the appropriate medical and allied health professionals are to assist us. Being able to meet with other members of our "CMT Family" in our area is very important. This provides the opportunity to learn from and be encouraged by others on their CMT journey.
One of the aims of the CMTAA is to provide the opportunity for people with CMT, or those caring for them, to speak with other members of our "CMT family". This can be done by:

a. Talking to one of our Regional Co-ordinators in your area. The CMTAA website www.cmt.org.au has a list of Support Groups throughout Australia with contact details for Regional Co-ordinators as well as links to credible CMT related organisations. To find a Support Group in your area, on the CMTAA website home page simply click onto "Current Activities" and on the scroll down list click onto "Support Groups".

b. Attending informative Awareness Day seminars and Support Group meetings that are advertised on our website and Facebook page.
c. Joining our Facebook page.
d. Contacting our National Office for further information – 02 9767 5105
Email - cmtaa2@cmt.org.au

Are other members of my family likely to have this and what impact will this have on my children?

There may be other members in your family who are obviously impacted by CMT. Some may know this, others may be in denial, and others may not have any symptoms at all. There are over 80 different types of CMT genes. The good news is that over 80% of CMT falls into 5 main genetic categories:

CMT Type 1A (PMP 22 gene)
CMT Type 1B
CMT Type 2
CMT Type 2A
CMT X Linked

We suggest that when you are told that you have CMT that you explore genetic testing ideally to establish what type of CMT you have.

This is important for future generations. To move down this path the first point of contact is your Neurologist who should be able to direct you to the appropriate organisation.
The question to ask your Neurologist: "*Can you suggest a Neurogeneticist or other appropriate person I can see to have a genetic test to determine what type of CMT I have?*"

Children have the benefit of a multi-disciplinary clinic located in Sydney where they can see neurological specific medical and allied health professionals during their appointment. This is:

The Institute for Neuroscience and Muscle Research,
The Children's Hospital
Westmead, Sydney NSW
Ph: 02 9845 1400

As a person with CMT we suggest that you do ask questions regarding treatments that may be prescribed for non-CMT situations. Specifically any adverse impact they may have on you due to your CMT.

Data informs us that 1 in 2500 people have CMT. This means that it is quite possible that the medical or allied health professional you visit may not have seen many patients (if any) who have CMT.

To assist you with your rapport, we suggest you ask the following initial questions:

1. *Do you know what Charcot Marie Tooth (CMT) disease is?*
2. *Have you previously seen a patient with CMT or patients with a peripheral neuropathy?*
3. *Are you aware of the CMTAA website www.cmt.org.au and links to other credible CMT sites?*
4. *As a person with a degenerative peripheral neuropathy, is the treatment/medication you prescribe going to impact my mobility and balance or be detrimental to my CMT?*
5. *I am determined to work at enhancing my quality of life and in managing my CMT. Can you suggest other medical and allied health professionals I can visit for further information that I can use to achieve this goal?*

Additional questions you may ask:

1. *What is available to enhance my balance?*
2. *Who do you suggest I see to help my mobility?*
3. *What can I do, whom can I see to ensure I am prolonging my quality of life?*
4. *What does Medicare contribute towards the cost?*
5. *Are there aids I can get to assist my daily living?*
6. *Do I qualify for NDIS assistance?*

We strongly suggest:

Keeping a folder so that you can take relevant notes every time you visit a medical and allied health professional. We can quickly forget what may have been mentioned at each visit. Additionally, prior to your visit, make a list of questions that you want to ask your medical and allied health professional.

Are researchers close to finding a cure?

There is significant worldwide research collaboration and progress is being made. The challenge is there are over 80 different types of CMT genes (and more genes are being discovered). Some types are close to having a medication trial however comprehensive testing needs to be carried out prior to getting the all clear to release.

In Australia we have leading researchers and their dedicated teams who are well respected for the world-class research they are carrying out. The CMTAA actively supports Australian research through our Research Grant programme and has contributed over $300,000 to this programme.

3. We Need To Manage Our CMT

Everyone is at a different stage on their CMT journey and therefore CMT management is specific to the individual. As a general starting point, lets look at the areas where CMT impacts the most.

Lower Legs and Feet

Situation	**Who to See**
Trips and falls	Physiotherapist; Exercise Physiologist; Podiatrist, Pedorthist
Muscle/ Functional core strength	Physiotherapist; Exercise Physiologist, Podiatrist, Occupational Therapist
Foot deformity,	Podiatrist, Pedorthist, Orthopaedics
Shoes that are comfortable	Companies that provide good quality broad shoes to accommodate orthotics and possibly AFOs
Musculoskeletal pain	Pain specialists
Nerve pain	Pain specialists

*After other options have been explored, we suggest that surgery be considered.

Exercise has been proven to be beneficial. Doing regular appropriate exercises including balance, stretching and strengthening exercises commensurate with the specific stage of your CMT journey can make a difference to your daily quality of life. Most importantly it is necessary to do prescribed exercises the right way and persevering with them to maximise the benefits. Appropriate mobility aids such as under toe supports, orthotics, and Ankle Foot Orthosis – abbreviated to AFOs can be beneficial.

A person with CMT may want/need to visit the following medical and allied health professionals:

GP
Neurologist
Neurogeneticist
Physiotherapist
Pedorthist
Podiatrist
Occupational Therapist
Clinical Psychologist
Dietician
Career Counsellor/Advisor
Rehabilitation Therapist
Genetic Counsellor
Exercise Physiologist
Orthopaedic Surgeon
Hand Surgeon
Pilates/Yoga

A Clinical Psychologist can give professional knowledge to equip you with the most appropriate mindset in managing your CMT.

We trust that you will find this information beneficial to assist you on your “CMT journey”.