Charcot-Marie-Tooth Disease (CMT)

Including 3 related types:

- Congenital Hypomyelinating Neuropathy
- Dejerine-Sottas disease
- Hereditary Motor and Sensory Neuropathy

Processing a new diagnosis

A diagnosis of CMT can be a huge shock for yourself, parents, siblings, extended family members and friends. It is normal to feel an overwhelming mix of grief, confusion, anxiety, loneliness and helplessness as your life has been forever changed. Everyone will have different ways of getting through this time but it's important to know that help and support are available for you, your child and family. Hear advice on processing your diagnosis from a community member with shared experience.

The video is available at **https://youtu.be/A9XM71knDVw.**

Getting help processing a new diagnosis

There are many ways you can seek support as you process a new diagnosis:

- A psychologist is a university-qualified health professional who can help you talk about your thoughts and feelings to understand and cope with the challenges you and your family are facing. Visit our page on **Psychology** for more information.
- A counsellor is a trained professional who can help you talk about and work through problems. Visit our page on **Counselling** for more information.
- A social worker can provide information and support to people experiencing a range of issues including family problems, anxiety, depression, crisis and trauma. Visit our page on **Social Work** for more information.
- A genetic counsellor can help you understand how CMT is inherited and whether there are any implications for other members of your family. Your specialist will be able to advise whether a genetic counsellor is required. Read more on our page about **Genetic Counselling**.
- Your state or territory neuromuscular organisation can provide support, advice and information about living with CMT. Read more on our page about **state and territory neuromuscular organisations**.
- Your GP can talk to you about a mental health plan and how you can use this to help you access the support and services you need. Read more on our page about **Wellbeing**.
- Other people and families living with CMT have also experienced the emotional rollercoaster that comes with a diagnosis of CMT and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with CMT in The Loop Community on our **Forum**.

How to talk to your child and others about a new diagnosis

Having a conversation with a loved one about a diagnosis of CMT is not easy. In fact, it will probably be one of the hardest talks you will have. But, like all difficult conversations, it is important and necessary.

There are many reasons why people may not want to have this conversation, mostly centred on wanting to protect their loved ones and themselves.

But avoiding the topic is not helpful. People may have noticed differences and changes that have occurred and/or heard the

condition being discussed.

So it is crucial that you have a role in your loved one learning about their/your diagnosis. This will enable you to support them as they process the information and to be on hand to answer any questions they may have. If you are the person with CMT it will provide you with support and someone to talk to. Together, you can learn about the journey ahead.

It will also allow you to:

- provide answers to questions in an age appropriate way
- correct any misinformation they may have heard or read
- show them that you have faith in their ability to handle difficult conversations.

There is no right time to start this conversation. Research suggests the earlier you talk to your loved ones about your/their condition, the more natural the conversation will become. The important part is to pick a time and commit to it.

Some important do's and don'ts:

- Do be positive but also realistic.
- Do reassure them that no one has done anything wrong and this is not their fault.
- Do tell them they/you will do many wonderful things in their/your life – they/you may just do them differently.
- Do tell the truth. Answer all their questions.
- Do consider your language. Try to avoid negative or emotive words, such as 'suffering from a condition'.
- Do use daily living examples that they can relate to.
- Don't avoid answering questions and don't shut the conversation down.

If you are having difficulty starting the conversation, try asking a couple of questions such as 'How are you feeling today?' or 'Do you know why you/they are having difficulty with your/their muscles?'

Remember, it's okay to be upset during the conversation but try to avoid breaking down as this will only cause greater distress. And if speaking with a child, remember children are resilient and they generally handle information of this nature far better than adults.

Understanding CMT and how it's diagnosed

Charcot-Marie-Tooth (CMT) is an umbrella term for a group of conditions that affect the motor (movement) and sensory (sensation) nerves throughout the body. CMT, also known as Hereditary Motor and Sensory Neuropathy (HMSN), was named after the 3 neurologists who first described the condition in 1886, Jean-Martin Charcot, Pierre Marie and Howard Tooth.

CMT is a genetic condition that people are born with but may not show symptoms until later in life. There are many different forms of CMT, with varying age of onset, severity and inheritance patterns, even for people with the same type of CMT. Over 30 different genes have been shown to cause the different types of CMT.

CMT affects about 1 in 2500 live births. This means there is about 10,000 Australians living with CMT and 3 million people worldwide.

What causes CMT?

CMT is a group of conditions that are caused by a change (mutation or fault) in a gene. The faulty genes responsible for CMT contain instructions for the production of particular proteins. These proteins are important for the functioning of the peripheral nerves (the nerves that travel from the brain and spinal column to the rest of your body). The genetic mutation affects the function of the peripheral nerves, meaning the messages between the brain and your arms and legs are disrupted.

Peripheral nerves are made up of 2 types of nerves:

- The motor nerves - control movement by sending messages from the spinal cord to the muscles
- Sensory nerves - carry pain and temperature messages from the hands and feet to the spinal cord.

When these nerves don't function properly, weakness and wasting of the muscles in the arms and legs occurs, usually below the knees and elbows. In some cases, the damaged sensory nerves reduces the ability to feel heat, cold, and pain.

Damage to peripheral nerves is also known as neuropathy. This is why CMT is also known as Hereditary Motor and Sensory

Neuropathy (HMSN).

What are the types of CMT?

There are over 100 different genetic types of CMT however over 80% of CMT falls into 5 main categories:

- CMT Type 1A
- CMT Type 1B
- CMT Type 2
- CMT Type 2A
- CMT X-linked

CMT Type 1 is the most common type and accounts for two thirds of all cases of CMT. Of this group, 60% have Type 1A.

The symptoms of Type 1 and 2 are similar but people living with Type 2 tend to have a wider range of age of onset of the condition and degree of disability.

Children with severe symptoms of CMT are sometimes classified as having CMT Type 3 – Congenital Hypomyelinating Neuropathy (CHN) also known as Dejerine-Sottas disease (DSD).

The classification of CMT into subtypes is complicated and there isn't a universal classification system used by all doctors around the world.

For more information on the different types of CMT see **Muscular Dystrophy Association 'Types of CMT'**

How is CMT diagnosed?

The road to diagnosis usually starts when a person begins to notice changes with their bodies, usually mild lower leg muscle weakness. This is known as the pre-diagnosis phase and for some people it can last years.

Seeking medical advice from your GP, who may refer you to a neurologist, as early as possible can help you manage the physical changes that are occurring and prevent complications.

The first step in diagnosis involves a neurologist taking a detailed patient history, including a family history. Your symptoms and a family history of muscle weakness, reduced reflexes and sensory loss can indicate CMT. People with CMT often have problems with their feet, therefore your neurologist will examine your feet to see if they are showing any signs of the foot problems associated with CMT.

Formal testing will then follow and may include:

- Genetic testing looking for changes in one of the genes that cause CMT. Testing for CMT1a (the most common type of CMT) may be done before NCS and EMG. If CMT1a genetic testing is negative, then EMG and NCS will be done before doing a neuromuscular panel, which looks at a number of genes causing nerve disorders. Further information about genetic testing can be found on the **Australian Genomics Health Alliance website**.
- Nerve conduction studies (NCS) – this involves placing electrodes (small sensors) on the skin and providing a small electrical stimulus to the nerve to measure the response. The motor (moving) nerves and sensory (feeling) nerves are measured. The speed and strength of the transmitted signals is measured. If the speed of the signal is slower than normal, this indicates CMT Type 1. With CMT Type 2, the speed of the nerve conduction is normal but the strength of the signal is reduced.
- Electromyography (EMG) is used rarely. EMG involves inserting a small needle electrode (sensor) through the skin to measure the electrical activity of the muscles.

Identifying the specific type of CMT can help the person living with the condition and provide additional information about whether further monitoring is required. For example, some types of CMT are at higher risk for hearing problems. However, arriving at this very specific diagnosis is not always straightforward because two people with very different symptoms could have errors in the same gene.

The genetics of CMT

Faults in four genes are largely responsible for the majority of CMT cases. These are:

- CMT1A (PMP22 gene duplication)
- CMT1X (GJBI gene mutation)
- CMT1B (MPZ gene mutation)
- HNPP (PMP22 gene deletion)
- CMT2A (MFN2 gene mutation).

Different types of CMT have different inheritance patterns (how it is passed from parents to children). CMT can be inherited via:

Autosomal dominant pattern

This is the most commonly inherited pattern for CMT with CMT Type 1 and some cases of CMT Type 2. All people have 2 copies of each gene – one inherited from each parent. In autosomal dominant inheritance, it only takes one of the faulty genes from one parent to pass on the condition. This single altered gene is sufficient to over-ride the normal functioning copy of the gene inherited from the other parent. Therefore, the chance of passing on an autosomal dominant condition to an offspring is 50% or 1 in 2.

X-linked pattern

CMT Type1X is inherited in an X-linked recessive pattern. The altered gene (GJB1) is located on the X-chromosome. Females have two X chromosomes, one inherited from each parent. In most cases females who inherit a faulty GJB1 gene will show no symptoms or only mild symptoms of the condition because the normal gene on the other X chromosome will compensate. Females with one altered GJB1 gene are known as carriers and they can pass the condition on to their sons. The daughters of carriers each have a 1 in 2 chance of being carriers.

Males have only one X chromosome, which they inherit from their mother, and one Y chromosome, which they inherit from their father. If a boy's mother is a carrier of an altered GJB1 gene there is a 1 in 2 chance that he will inherit this faulty gene. If so, the boy will have CMT because he doesn't have another X chromosome to over-ride the faulty one.

Autosomal recessive pattern

Less commonly, some CMT Type 2 cases are inherited in an autosomal recessive pattern. This means an individual needs two faulty genes – one from each parent. The parents of an individual with this type of CMT each carry a copy of the altered gene. They are known as 'carriers' and typically do not have symptoms of the condition as the other 'normal' copy of the gene is enough to prevent the condition developing . For carrier parents to have a child with LGMD Type 2, both parents must pass the altered gene on to their child. If both parents are carriers the likelihood of a child inheriting the condition is 25% or 1 in 4.

For more information on the inheritance of specific types of CMT, see Muscular Dystrophy Association '**Causes/Inheritance of CMT**'

Long-term outlook

The impact and progression of CMT varies from person to person, even amongst people with the same type of CMT. Some people have little difficulty throughout their lives while other people will required greater assistance.

Most people with CMT are able to continue walking, though sometimes with mobility aids, and have a normal life expectancy.

Understanding and planning for changes

Learning about CMT and surrounding yourself with the right healthcare providers, services and support can help you feel more in control of what lies ahead. Although every person's journey with CMT will be unique, the information below will help you understand this journey and how to prepare and plan for changes in the future.

What are the main symptoms of CMT?

The symptoms of CMT, and severity of symptoms, vary greatly amongst people. In the most common types of CMT, symptoms usually begin before the age of 20 years. The typical symptoms of CMT are:

- Weakness and wasting of the muscles of the lower legs and feet – this is often the first sign and presents as clumsiness, tripping and falling.

- High arches in the feet (known medically as pes cavus) and curled toes (hammer toes) – can lead to instability of the foot and ankle, increasing the risk of tripping and ankles injuries
- Foot drop (inability to hold foot horizontal when foot is lifted off the ground) – causes people to walk with an awkward high stepping gate, increasing the risk of tripping
- Numbness in the feet and legs – if numbness is severe, your feet or legs can get injured without you realising. Daily checks of the skin on your toes and feet are important if you experience numbness.
- Difficulty with balance
- Hand weakness and numbness, often appearing after leg problems have commenced. Some people with CMT also experience hand tremors.
- Fatigue as a result of the extra effort required to perform daily activities.

Some very rare symptoms of CMT include curvature of the spine (scoliosis) and speech, swallowing and breathing difficulties, particularly when lying flat at night.

Progression and monitoring

Most people with CMT remain able to walk throughout their lives. As the condition progresses they may require walking aids and sometimes a manual wheelchair for occasional use. It is rare for a person with CMT to need to use a wheelchair fulltime.

As the condition progresses you might also notice:

- Hand and forearm muscle weakness – this can lead to the loss of fine motor control, dexterity, and overall hand strength making it difficult to complete daily activities such as doing up buttons, typing, holding a pen and opening jars.
- Pain – this is often associated with poor walking technique and the stress and strain that it places on the muscles and joints. More rarely the nerves themselves cause pain know as neuropathic pain.
- Tremors (muscle shaking) – some people experience tremors in their arms and legs. Severe tremors are called Roussy-Levy Syndrome.

Symptoms often progress noticeably at the time of a growth spurt, particularly during puberty.

Community Advice

Hear from a community member who has walked the path before you.

The video is available at **https://youtu.be/I7labxjWv9w.**

Life stuff

To find out more about living life with a neuromuscular condition and to access stories and peer-advice from the community, visit our **Living Life** section.

Where to find more information about living with CMT

Charcot-Marie-Tooth Association Australia
Provides support and information to the CMT Australian community.

Charcot-Marie-Tooth fact sheet,
From Muscular Dystrophy Association Australia, June 2018

Charcot-Marie-Tooth fact sheet
From Muscular Dystrophy New South Wales, 2017. This fact sheet includes considerations for future planning.

International Organisations

Charcot-Marie-Tooth Association USA

Charcot-Marie-Tooth UK

Muscular Dystrophy Association USA

Treat-NMD

Support for CMT

Treatment and Management

At the moment there is no cure for CMT but there is a lot you can do to improve your quality of life. Being a rare disease, it is often not well understood by the general medical community and you may find you need to work in partnership with, and help educate, your healthcare team.

Feet

Many people with CMT have foot problems, from the structural changes and reduced skin sensation in their feet. It is important to keep your feet in good health and prevent potential problems with treatments including:

- Stretching of the feet to reduce pain
- Correct fitting orthoses (shoe inserts) to help spread the load on the foot, reduce pain and prevent further foot damage
- Keeping feet warm as CMT can reduce circulation
- Daily checking of the feet is important especially for people experiencing reduced foot sensation and numbness. Seek help from a podiatrist, doctor or nurse to remove calluses and corns to prevent accidental injury to the skin.
- See a podiatrist to assist with foot care management, as well as advice about shoes and orthotics (shoe inserts).
- Correct footwear is essential to reduce the risk of falls and tripping but can be difficult to find for people with high arches and/or hammer toes
- Applying skin cream to the feet, except between the toes, to prevent foot dehydration, skin cracking and itching
- Correct-fitting and clean socks to avoid skin infections
- Foot surgery to correct foot deformities is a last resort, as success is variable.
- Exercise regularly
- Get enough sleep
- Keep well hydrated by drinking plenty of water
- Maintain a healthy weight
- Reduce stress
- Reduce caffeine intake
- Reduce alcohol consumption.

To read more, visit our section on **Podiatry**.

Hands

Treatment and management of problems associated with the hands should focus on working with an **occupational therapist** and include;

- Stretching – daily stretching may reduce pain and joint deformity. Focus should be on the wrists and fingers but should never be painful.
- Exercises – in some people exercises for the hands and wrists may help with maintaining strength and preventing muscle wasting. Activities that engage the hands in fine motor, coordinated actions may also help. This can include activities such as playing a musical instrument, playing games like jenga or doing needlepoint.
- Splinting – an OT may decide that a splint is required to increase hand function or to prevent hand deformities.
- Assistive technology – sometimes your OT may need to look at modifying a task or the use of assistive technology, which can help you with daily living tasks and maintaining independence.

Falls

Symptoms such as muscle weakness and reduced sensation can increase the risk of falling. Falls can be dangerous for

people with CMT as broken bones take longer to heal and inactivity while recovering from a fall may worsen muscle weakness. Good shoes, clearing any trip hazards from your home and taking care when walking, particularly on uneven ground, can all help to avoid falls. Seek advice from a physiotherapist or occupational therapist to learn how you can reduce your risk of falling at home and when out and about.

Visit our pages on **Physiotherapy** and **Occupational Therapy** for more information.

Fatigue

People with CMT experience fatigue as a result of the extra effort required to perform daily activities. An occupational therapist can provide advice on energy conservation strategies. Other strategies listed by the Muscular Dystrophy Association (US) include:

- exercise regularly
- get enough sleep
- keep well hydrated by drinking plenty of water
- maintain a healthy weight
- reduce stress
- reduce caffeine intake
- reduce alcohol consumption.

Visit our page on **Occupational Therapy** for more information.

Physiotherapy and exercise

Regular balance and stretching exercises that are tailored to your specific stage of CMT can maintain your general health, movement, muscle strength and flexibility. Moderate activity is usually recommended and over-exertion should be avoided. Many people with CMT find swimming or hydrotherapy to be an excellent form of exercise. A physiotherapist can work with you to design an exercise program that is safe and helps you achieve your goals.

Exercise can also assist with maintaining a healthy weight. This is very important for people with CMT as it reduced stress on already compromised joints and muscles.

Visit our pages on **Physiotherapy** or **Hydrotherapy** for more information.

Pain and stress

Living with pain and high levels of stress can have a huge impact on your daily life and your overall wellbeing. There are many things you can do to help manage the pain and stress you might be experiencing. Speak to your health care team or a pain specialist about options for the management of your pain, including medications, exercise, equipment/devices and learning to change how you think about pain. There are many sources of support you can turn to when you are feeling down or needing some extra help to manage stress or other feelings, including state and territory neuromuscular organisations and other community organisations. You can also seek support from your doctor, a psychologist, counsellor or social worker.

Visit our section on **Wellbeing** for more information.

Breathing problems

A small percentage of people with severe CMT have problems with their breathing, usually at night. Signs include frequent chest infections, daytime sleepiness and morning headaches. People experiencing these symptoms should consider a referral for a sleep study. This may lead to the use of a non-invasive ventilator device at night. People with CMT who experience breathing problems should also talk to their doctor about the flu and pneumococcal vaccines. You can find out more by visiting our **Respiratory Care** page.

Source:

Charcot-Marie-Tooth
Muscular Dystrophy Australia

Moving Forward with CMT
Charcot Marie Tooth Association Australia

Other CMT Care Guidelines

Why Feet Matter
From Charcot Marie Tooth Association Australia

A Guide to Physical and Occupational Therapy for CMT
From Charcot Marie Tooth Association (US)

Family planning

Family planning options are available to families where the CMT type has been identified. If the genetic variant has been identified in the family, it is possible to perform testing to determine whether a baby will have the condition. See your GP or a genetic counsellor for more information.

To read more, visit our page on **Genetic Counselling**.

Other helpful support services

The video is available at **https://youtu.be/BDaCNsd9R_s.**

- Your **state or territory neuromuscular organisation** can provide information and advice on what support they can offer such as local support groups, camps, programs, services in the local area, access to cough assist machines, advocacy or assistance in times of crisis. They can also provide an ear to listen if you need someone to talk to or guide you to get the assistance you are needing.
- Seeing a psychologist, counsellor or social worker can be incredibly helpful if you, your child or other members of the family are having a tough time or struggling with negative thoughts and feelings. Learn more about what services are available and how to access them here: **Psychology**, **Counselling**, **Social Work** and **Wellbeing**.
- Living with CMT may mean that you or your child may need some assistance for everyday activities. Find out more about how a disability support worker could help and how to engage this type of support. Visit our page on **Disability Support Workers** for more information.
- Other people and families living with CMT have also experienced the emotional rollercoaster that comes with a diagnosis of CMT and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with CMT in The Loop Community on our **Forum**.

Medical Alert Card

Muscular Dystrophy Queensland has developed a **Medical Alert Card for CMT**. A Medical Alert Card can help you communicate your care needs in a medical emergency situation. If you would like a Medical Alert Card to keep in your wallet or purse, please call (07) 3243 9700 or complete the form on the Muscular Dystrophy Queensland's **website**.

How to have better conversations when communicating your needs

To learn how to have better conversations when communicating your needs, visit the following pages:

Employers

Educators

Living Life: Education

About CMT

Charcot-Marie-Tooth fact sheet
From Muscular Dystrophy Association Australia, June 2018.

Charcot-Marie-Tooth fact sheet
From Muscular Dystrophy New South Wales, 2017. This fact sheet includes considerations for future planning.

Charcot-Marie-Tooth
Muscular Dystrophy Australia.

Moving Forward with CMT

Charcot Marie Tooth Association Australia.

CMT Fact Sheet

From National Institute of Neurological Disorders and Stroke (United States).

Charcot Marie Tooth Disease

This short animated film was made for the Rare Film Festival 2020 by students studying at the University of Greenwich. Shortlisted in the category: Best Aspiring Filmmaker - Charity Collaboration.

CMT Care Guidelines

Why Feet Matter
From Charcot Marie Tooth Association Australia.

A Guide to Physical and Occupational Therapy for CMT
From Charcot Marie Tooth Association (US).

Therapies for Inherited Neuropathies

From Charcot Marie Tooth Association (US). A comprehensive fitness program for all levels and abilities that focuses on the following five areas: Flexibility, Balance, Strengthening, Endurance and Function. Developed by CMTA Board Member Steve O'Donnell in partnership with renowned Physical Therapist/Neurologic Clinical Specialist Mike Studer.

Support Organisation

Charcot-Marie-Tooth Association Australia Inc

The CMT Association Australia Inc. (CMTAA) began as a Support Group in 1988, and was incorporated in 1990 as a registered charity. The motivation to establish the CMTAA Inc was to meet the need for people with CMT to communicate with each other and provide mutual encouragement. Over the years we have slowly "evolved" and are now moving forward at an unprecedented pace.

International Organisations

Charcot-Marie-Tooth Association USA

Charcot-Marie-Tooth UK

Muscular Dystrophy Association USA

Treat-NMD