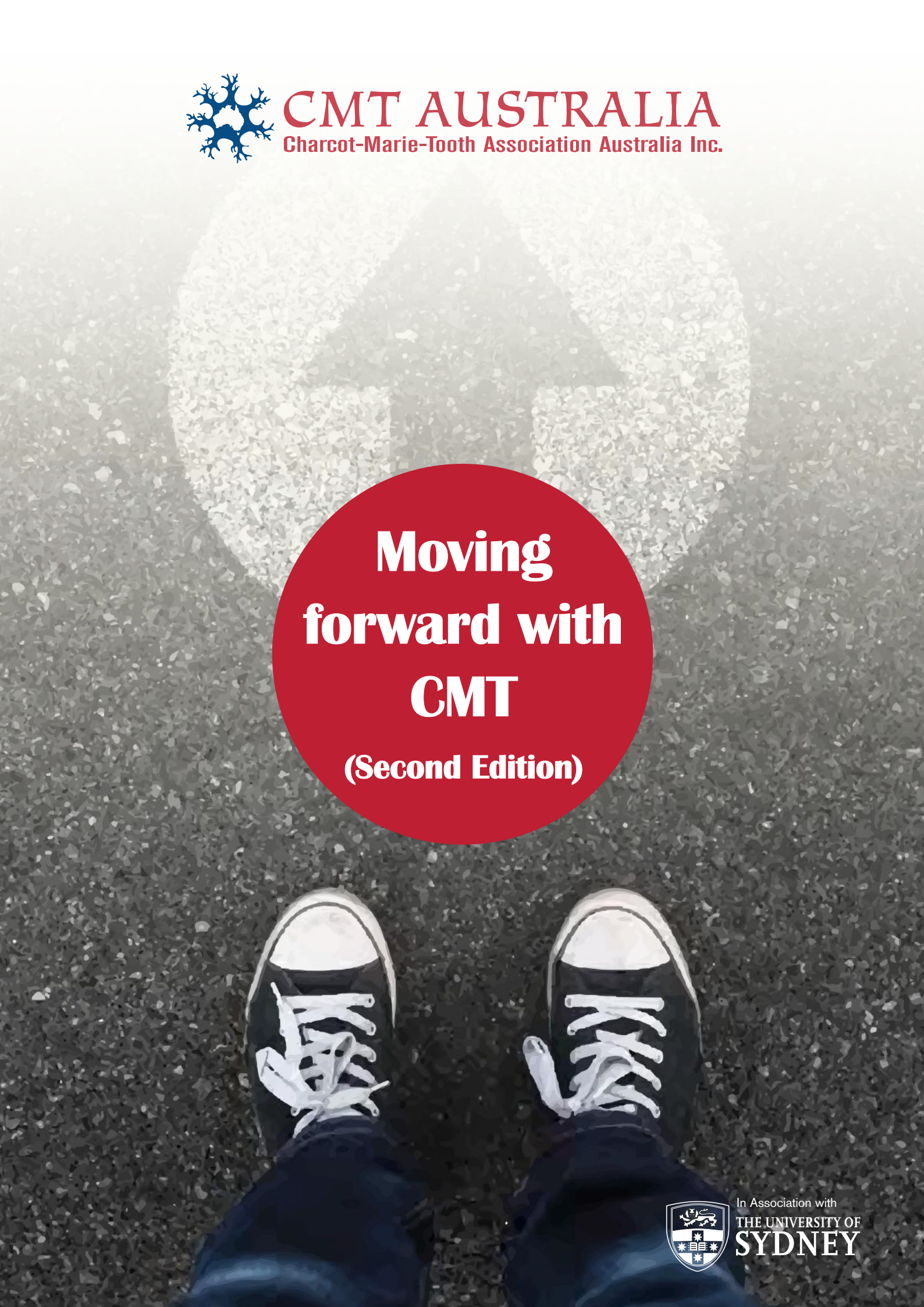
CMT AUSTRALIA
Charcot-Marie-Tooth Association Australia Inc.

Moving forward with CMT

(Second Edition)



In Association with
THE UNIVERSITY OF SYDNEY

Welcome

The Charcot-Marie-Tooth Association of Australia Inc. (CMT Australia) welcomes you to our community. A diagnosis of Charcot-Marie-Tooth (CMT) may be unsettling with information provided overwhelming. his booklet is offered as an introductory guide, designed to offer you an overview of CMT after diagnosis. More specific information is available to members via our newsletter, national office, website (www.cmt.org.au), by attending online or in person any of our popular Annual National Conference, and/or support group meetings.

As President of 'CMT Australia' it is my pleasure to introduce you to our second edition of 'Moving Forward with CMT'. This informative document was first published in 2016, and since then has served as an important introduction to many as they navigate their journey with CMT. Dr Scott Denton PhD compiled our initial edition, and I'm pleased to say that Scott has again contributed to this update. Seven years on, there is still no cure for CMT; however, through research taking place both in Australia and internationally, the vision we hold of **'a world without CMT'** is now appearing on the horizon.

Our objective here is to collect into one publication insights into the many aspects of Charcot-Marie-Tooth disease (CMT). There are articles ranging from an introduction to how the name originated, through to the 'latest research. We review the symptoms, ways of diagnosis, who you can turn to for advice, and what CMT Australia is doing to keep you up-to-date and 'moving forward'.

Ultimately, we are all responsible for managing our CMT, and this is aided by publications such as this one. I encourage you to explore its content and to join our Association so that we can share our experiences and strategies through the opportunities such an Association provides.

Robert Twin

President CMT Australia

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CMT AUSTRALIA
Charcot-Marie-Tooth Association Australia Inc.

What is CMT?

Charcot-Marie-Tooth (CMT) is a strange name for a disease, but it is the most common inherited peripheral neuropathy. CMT is named after Jean-Martin Charcot, Pierre Marie, and Howard Tooth — three European physicians who first identified the condition in 1886. CMT is also known by another name; Hereditary Motor and Sensory Neuropathy (HMSN). With an estimated occurrence of one in every 2,500, CMT affects around 10 000 Australians and 3 million people worldwide.

CMT is a genetic disease that damages the peripheral nerves that send brain messages to the muscles (motor nerves). Peripheral nerves also send messages back to the brain to do with sensations (sensory nerves) like touch, hot or cold, pain and balance. When the sensory nerves that tell the brain where your joints are in space are damaged, it is called a neuropathy, thus CMT is a neuropathy. Importantly, CMT is rarely life threatening.

Due to damaged nerves, people with CMT over time may exhibit reduced sensation and slowly weakening muscles, predominantly in feet and hands. The severity, or how far or fast this will progress, varies extensively from person to person. Some go about their lives with little difficulty, while others require greater assistance. CMT affects people differently, even in the same family.

CMT is really an 'umbrella term' for a range of certain inherited genetic conditions that affect peripheral nervous system. Accordingly, CMT is divided into types and subtypes. Currently, over 100 different genes causing CMT have been identified and as geneticists discover further variants, the list continues to grow. CMT Type 1 is the most common type of CMT, accounting for about two-thirds of all cases. A subtype of CMT Type 1, called CMT 1A accounts for approximately 60% of CMT Type 1 cases, making it the most common CMT subtype.

Although there is currently no cure for CMT, there are many successful treatments both in place and under development, which may be used to effectively manage symptoms. Extensive ongoing research worldwide is encouraging and includes efforts to improve diagnosis, offer preventative measures, as well as slow down or even reverse nerve degeneration and muscle atrophy.


Jean-Martin Charcot


Pierre Marie


Howard Henry Tooth

CMT
Diagnosis and beyond

Diagnosis is a significant part of your CMT journey. The vast majority of people knew something was amiss with their bodies before diagnosis. In this pre-diagnosis period, for some spanning decades, people may often face a range of emotional and physical challenges.

Early diagnosis often enhances quality of life by treating and managing physical changes resulting from CMT, as well as assisting with mental health issues often associated with CMT such as anxiety and depression. The age of diagnosis can cover the full spectrum of life, from birth to retirement. Some people can live almost their entire life without knowing that they have the disease.

Initial Symptoms

Since CMT is mostly hereditary, it is important to tell your neurologist about family members with CMT symptoms. It is often a family member who first notices symptoms of CMT, which include lower leg weakness, clumsiness, foot and hand deformities.

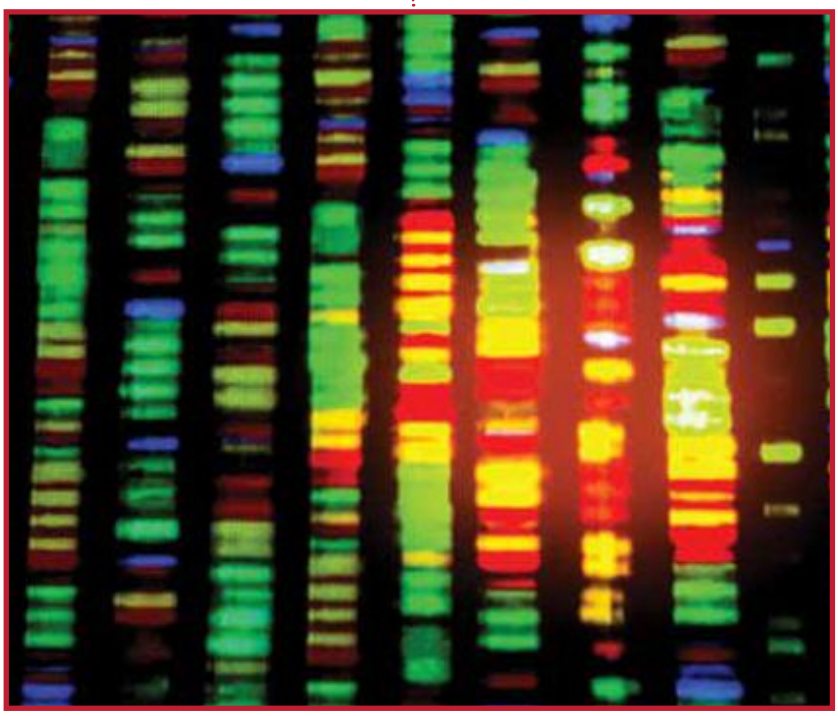
People may exhibit 'champagne legs', where the leg is very narrow at the ankle, but widens in the upper thigh, giving the appearance of an inverted champagne bottle. Other typical features include high arched feet, foot drop and a high stepped gait. Many have difficulty with slippery and uneven surfaces and often fall, especially in the dark. These are some of the first signs that something is wrong.

Formal Diagnosis

In Australia, 77% are first told they had CMT by a neurologist, with 6% informed by their GP. Due to the hereditary nature of CMT, it is often a family member (usually a parent) who informs the individual. No family history of CMT does not exclude the possibility of having the disease. This is known as a 'new mutation' or 'sporadic' case of CMT.

Formal testing by a neurologist may be electro-diagnostic and/or genetic. Electro-diagnostic tests are commonly used to diagnose CMT through nerve conduction velocity, which calculates the strength and speed of electrical signals travelling along the peripheral nerves. Genetic tests, done by taking blood, check for many, but not all, common chromosomal defects causing CMT.

Most people who have had a genetic test have one of five conditions, caused by mutations in four genes: CMT1A (PMP22 gene duplication), CMT1X (GJB1 gene mutation), CMT1B (MPZ gene mutation), HNPP (PMP22 gene deletion), or CMT2A (MFN2 gene mutation). A positive genetic test will provide conclusive diagnosis.



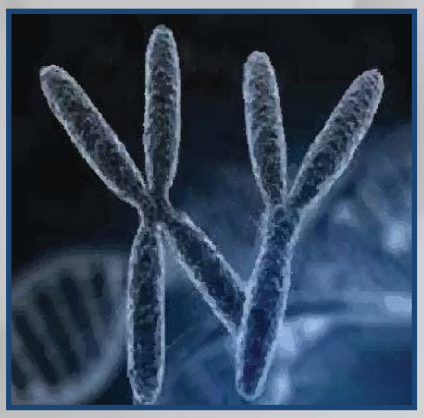
Family Planning

Once a diagnosis of CMT has been made in the family, it is natural to start thinking about the chance another family member may have the condition. For couples who have not completed their families, their thoughts may be directed at the chance that a future child may inherit CMT. Family planning options may be 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 be affected with this condition. Some families may choose this pathway to better prepare themselves before a baby is born, or to ensure that any future children will not be affected with CMT. Other families may elect to fall pregnant without testing and wait until the baby is born to be checked for symptoms of CMT. There is no right or wrong way when planning a family: it is an individual choice, and your doctor will support you through whatever options you choose.

Pre-Implantation Genetic Diagnosis (PGD): is a specialised technique where embryos created via invitro-fertilization (IVF) are tested for the CMT gene mutation in your family before they are implanted into the mother's uterus. Only those embryos that do not carry the CMT gene mutation are transferred, ensuring that the resulting baby will not be affected with CMT.

For further information about the implications of PGD, please consult your doctor or genetic counsellor. If you would like more information about all family planning options, you may contact your local genetics service. A referral from your family doctor will be required. For more information and locations of genetic services in Australia, please visit the Centre for Genetics Education website: www.genetics.edu.au.



Medication safety and CMT

From time to time we all need to take medication, and it is to be expected that you will need to take medications for conditions not related to your CMT. Sometimes the side effects listed for a medication can sound alarming. The good news is that for most medications, people with CMT are generally at no greater risk of side-effects than the general public. Two exceptions include the cancer drugs Vincristine and Paclitaxel, where scientific evidence shows drug-related worsening of CMT.

If you are concerned how a certain medication might impact on you, free resource materials to assess medicine safety is available, from NPS MedicineWise online www.nps.org.au or phone 1300 MEDICINE (1300 633 424) to speak with a registered pharmacist. It is recommended to consult your pharmacist, family doctor, or neurologist about any concerns so they can answer your questions and actively investigate medication safety that is specific for you.

Most practitioners will talk about 'risk' and will often not say a medicine is 'safe', because 'safe' is open to interpretation and means different things to different people. Health practitioners assess risk of medications against the benefit to your condition, for example it is better to treat your cancer even if medications used might impact on your CMT. There are often multiple options available to treat a particular condition so there may be alternative medications if you are not comfortable with the level of risk presented to you. Your pharmacist may also offer assistance to help you to understand medicine information and make recommendations to you (and your doctor) about how best to manage your medical conditions.

Finally if you believe that you have experienced worsening of your CMT as a result of taking a medication, then you should seek advice from a medical professional. You can also report this adverse event to the Therapeutic Goods Administration online at www.aems.tga.gov.au or by phoning the Adverse Medicines Events (AME) line (1300 134 237). This can be done personally or in consultation with your health practitioner.

CMT
and your feet

Preventing and treating foot problems related to CMT

High Arches (pes cavus)

The classic, very high arch is formed because the peroneal group no longer properly opposes the tibial muscles. Muscles need to be balanced. The tibial muscles get "free reign" and work without an opposing balance. This results in a pulled-up arch, tipping the foot outwards, which creates instability, and tiredness and can lead to ankle sprains.

Treatment may range from bracing with orthosis, deep shoes with soft inlays or surgical realignment. Regular stretching and exercise are recommended to maintain flexibility and strength.

Hammer Toes

Hammer Toes (clawed toes) are caused when the muscles within the foot itself (intrinsic muscles) weaken and cause the toes to curl up. The contracted toes do not have enough room in shoes and put increased pressure on the ball of the foot, which becomes painful and callused.

Treatments include toe splints, stretching out the toes and insoles. Make sure to wear shoes that have a wide toe box to minimize pressure on the toes. A custom orthotic can help relieve pressure on the ball of the foot and surgery can also straighten the toes.

People affected by CMT often display distinct physical symptoms that manifest in their feet and foot care is at the forefront of their management. Here are the most common foot symptoms and how to treat them.

It is strongly advisable for people with CMT to see a podiatrist, preferably one who has experience with CMT.

Paediatric podiatrist Professor Joshua Burns PhD treating James Houghton at the Peripheral Neuropathy Management Clinic (PNMC) based at The Children's Hospital at Westmead. The PNMC has touched the lives of many children and families with affected muscle, nerve, and brain, giving them hope for a healthier future. The Children's Hospital attracts the best new researchers from Australia, New Zealand, and Europe to lead the challenge to develop therapies to better the lives of Australian children.

Fungal Toenails

People with CMT are also prone to fungal foot infections, which can make nails thick and painful. Preventative solutions such as correct socks can avoid secondary bacterial infections that require antibiotic treatment.

Exercise, proper hygiene and regularly inspect your feet and toes. See your family doctor or podiatrist rather than cut corns, calluses or ingrown nails yourself.

"High-arched feet (Pes cavus) is a very common characteristic."

Foot Dehydration

CMT can cause foot dehydration due to low natural lubrication.

Skin cracks may induce severe itching and scratching that can lead to openings that may become infected.

Skin cream on every part of the foot except between the toes can assist.

Foot Drop Gait

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).

To reduce the instances of tripping due to foot drop, carbon fibre lightweight ankle-foot othosis (AFOs) that fit into most shoes and are easy to fit around the calf show considerable success. While effective treatments include AFOs and rocker bottom-style shoes, in some instances surgical transfer of stronger muscles to the top of the foot can also help stabilise foot drop.

"Hammer toes (clawed toes) and corns on the toes are usual."

Sensory Loss

Sensory loss can be potentially dangerous. This is because ulcers can occur without pain. Frequent inspection of your feet is essential.

Orthotists - supporting you

The most common orthotic treatment for CMT is to manage foot deformities and drop foot by using functional insoles and AFO's. Orthoses are designed to be placed in the shoe to help counteract the effects of CMT by keeping people mobile while minimising pressure on other parts of the body. Types of orthoses range from simple insoles to lightweight carbon fibre splints and custom-made footwear. People with CMT have different needs and your orthotist will insure that you receive the right support for you. Physiotherapist's can also help with making new orthosis comfortable.

Foot surgery

While foot surgery is at times necessary for people with CMT, it is recommended to be a last resort. Surgery focuses on bone and repositioning the high arch, ankle laxity and tendon transfer. Australian CMT studies show mixed results with surgical procedures to the feet. Surgery has risks, whether it's from the surgery itself, anaesthetic or the recovery period.

CMT Aussie Kids

"I met so many great people who know how I feel and the struggles I face."

"It's always awesome to meet people that go through the same challenges."

"It was really good to meet other kids my age with CMT."

"The camp pushes me to achieve things I never thought I could."

CMT Australia runs a program for youth living with CMT in Australia and New Zealand.

The program includes the annual CMT Aussie Kids camp, Family Fun Days, CMT Big Kids, community activities, online virtual get togethers and social media.

Most kids had not met other kids with CMT before. The camp offers an opportunity to meet other kids with CMT around their own age in a safe, supporting environment.

Adult volunteer leaders have CMT or are closely related to someone with CMT.

The Family Fun Days help bring together our youngest Aussie Kids and their families at local attractions.

Targeted at kids too young for camp, the Family Fun Days also provide a great social outlet for young CMT families.

CMT Aussie Kids National Camp

CMT Aussie Kids, an activity of CMT Australia, runs a national camp exclusively for youth living with CMT. CMT Aussie Kids Camps are for youth aged 8 - 18 from across Australia and New Zealand. 'Kids' can meet with their peers to share the good times and the stresses, joys, and successes, explore, and enjoy challenging and adventurous activities in a safe environment; and learn about CMT.

Activities vary and may include canoeing, sailing, indoor rock climbing or surfing.

Beginning in 2012 with 11 kids, the camp more than doubled in size in its first four years and now boasts attendees from across Australia and New Zealand. One of only three such camps in the world, the camp enjoys close relations with its UK counterpart including reciprocal visits.

We encourage young adults as 'Mentors' to act as role models to the newer attendees, further developing a sense of achievement and responsibility as well as positioning our CMT youth for future roles in the community.

Interested? For further information contact CMT Australia or email cmtaussiekids@gmail.com.

A further addition to the camp, CMT Aussie Kids Facebook and Instagram pages allow youth living with CMT to connect and support each other through social media.

Visits to CMT research labs further extend our community outreach activities.

Activities catering for the 'Big Kids', young adults aged 18-30, living with CMT.

Driven by the attendees themselves, the activities reflect their own interests and diverse skills while they support one another and share friendship.

CMT Aussie Kids
www.cmt.org.au
Phone: 0458 630 631 Email: info@cmt.org.au

CMT Australia
Benefits of membership

AU $40.00 /yr.

The CMT Association Australia Inc. (CMTAA) began as an informal support group in 1988, incorporated in 1990 as a registered charity, adopting the public name of CMT Australia in 2018. CMT Australia was established as a means for people with CMT to have the ability to communicate with each other and provide mutual encouragement. Since these beginnings, CMT Australia's charter has grown to enhance the quality of life for people with CMT to:

1. Keep members informed of new developments in research, management, and all things CMT through:
   a. National office
   b. Website www.cmt.org.au
   c. Regular newsletter
   d. Facebook
   e. Interactive seminars
2. Actively encourage and support CMT research
3. Actively promote and encourage local and regional support groups
4. Link youth with CMT through supervised annual camps, day activities and social media
5. Promote CMT awareness to medical and allied health professionals
6. Promote CMT awareness to all levels of Government
7. Promote the development of facilities and services for people with CMT
8. Provide simple online membership and donation opportunities
9. Encourage members who believe they have the skills and passion to contribute to the growth and development of CMT Australia to join our management team.

"After attending just one CMT Australia seminar I gained the information to improve my mobility immensely. The regular newsletters keep me up to date with the latest research."

Dr Scott Denton PhD, a person with lived experience of CMT speaking to findings from his Department of Health study 'Reducing the Health Burden of CMT in Australia' at the 2015 CMTAA Annual Seminar held at Concord Repatriation Hospital.

CMT Community

The Critchley family

The Critchley family; Peter, Jillian, Matilda, and Eleanor are dedicated long-term members of CMT Australia. The Critchley's have appeared on ABC's science program Catalyst, SBS' Insight, in mainstream media, as well as CMT Australia seminars. Peter, Matilda, and Eleanor have CMT 1A.

In 2012 Peter and Jillian founded 'CMT Aussie Kids' and are active CMT Australia Committee members. Peter completed his PhD in Engineering in 2023 and Jillian is editor of the CMT CoMmuniTy Newsletter. Both actively participate in CMT research as patient advocates and investigators.

At the age of 8, Matilda was diagnosed with hip dysplasia, a condition frequently associated with CMT. After significant surgery, Matilda spent over two months in bed unable to sit or stand followed by two months in a wheelchair with a 'broomstick plaster'. Taking a positive approach to this difficult experience, Matilda developed a passion in health care and has since become a Registered nurse.

Eleanor has patiently participated in many clinical trials, including Botox and Vitamin C, and a range of interventions to address the personal impact of CMT including night splinting, serial casting and AFOs. She is studying Law and Psychology with a vision to work in disability advocacy.

Matilda and Eleanor are an integral part of the development of the CMT Aussie Kids program, providing support, encouragement and understanding, mentoring new members who may feel anxious meeting others with the same condition. They have been instrumental in the development of the 'Big Kids' Program together with other Mentors which will ensure the long-term future of the CMT Aussie Kids.

CMT Australia Area and State Coordinators

Research tells us that having a strong support network has many positive benefits. For people recently diagnosed with a health condition, some of the best benefits include higher levels of well-being, better coping skills, and a healthier life.

CMT Australia has a network of dedicated and friendly volunteers throughout Australia. These state and regional coordinators offer you the opportunity to contact someone in your area with a lived experience understanding of CMT.

You may just wish to touch base and talk with someone one-on-one, or you may wish to connect with a small group for a coffee and a chat. Regardless of how you connect, it is important to know that you are not alone.

CMT Australia support groups strive to exchange practical information related to CMT, where you can share similar experiences and local knowledge of services in your area. To contact your appropriate state and regional coordinator, scan the QR code or go to cmt.org.au and click 'Contact Us' on the Home Page.

CMT
Regional Australia

Nickolas Peachey

Nick's journey began in his early teens when he was diagnosed with CMT. Although the diagnosis was tough it answered a lot of questions. Through his many surgeries Nick began to get a better understanding of his CMT and how to live life positively with a disability. Although these have been some of the toughest years of Nick's young life, he is giving back to the CMT community that provided the love and support he needed. Initially joining the CMT Aussie Kids he now volunteers as a Mentor and Big Kid and is a member of the CMT Australia Committee and is training to become a professional speaker to inspire others to help themselves.

Kylie McIntyre

Growing up in Deniliquin Kylie's family had funny legs and tripped a lot. This didn't stop them doing anything. It wasn't until the late 1980's that the Thomas family found out there was a condition called CMT and all those dodgy feet and knees could be attributed to this. Kylie was involved in everything and from her multiple visits to the Radiology department as a child decided to be become a Radiographer. This allowed Kylie to be a part of the medical world and be very involved in research. Sarah and Thomas were born, and both tested positive for CMT. This just made Kylie more involved in the CMT community and a part of CMT Australia.

Roger MacRury

Not only can Roger MacRury fly a plane, he can build them too. Roger is a long-term resident of Canberra and hangers the plane he co-built in Goulburn. A very well-respected member of CMT Australia, Roger is the current Support Coordinator for the Australian Capital Territory. Roger enjoys flying as it gets him places around Australia, including flying to visit family members in Melbourne. Roger was not formally diagnosed as having CMT until he was 45, although the symptoms were evident from his early childhood. Now retired in his early 70's, Roger has enjoyed a long career in the Commonwealth Public Service for the Bureau of Meteorology, Treasury, Health and Defence.

The Douglas Legacy A hereditary journey

Phyllis Critchley (nee Douglas) has made long and significant contributions as a research subject and founding member of the CMTAA. This is Phyllis's story in her own words.

From my early years as a child living in the 1940's I knew I had inherited my dad's funny feet. I suffered a lot with painful legs and had difficulty running and walking. Back then, my doctors thought it was just growing pains, even though I had a brother and sister with the same problem. Growing up was difficult as I couldn't do what came naturally to others. I was made fun of because I was unable to jump the skipping rope or climb trees.

The years passed and in 1967 I was living in the USA, married with twin boys, Peter and David and a daughter Melinda. The boys were 18 months old and walking on their toes. I took them to an orthopaedic specialist and told him I thought they had inherited my feet; he didn't agree and thought the problem was due to my small build and that the twins were cramped in my womb which twisted their feet. We now know that this diagnosis was wrong.

In 1969 I accompanied my sister to Randwick Hospital after an operation on her feet was not responding as expected. We were both diagnosed with CMT and told it was rare, thus began our CMT journey. During the 1970's I participated in studies but lack of funds led to premature end.

Early in the 1980's Dr Nicholson from Concord Hospital contacted me and requested my involvement with his CMT research; he asked me how many extended family members had 'funny feet'. I couldn't even guess, as my father was one of nine children and his grandparents had sixteen children, and his great-grandparents also had a family of sixteen.

John Douglas, Phyllis's Great-Grandfather (left).

For six months I searched for related families I had never met. Dr Nicholson needed both CMT and non-CMT people to be tested to help isolate the genes. After six months I had contacted more than 200 people directly related to me for testing.

During my search I found relatives living in my street and another family only three streets away who had photos of my great-grandparents and an article about my great-grandmother being a midwife who had 16 kids and 63 grand kids.

In 1988 I read a notice in the paper of a CMT support group. Here I met Anycie Berkmann, Lyla Coorey, Kay Boreham, Elizabeth McDonald, and David Fennell. It was this group that formed the beginnings of the CMTAA. It was good to meet others with CMT and in 1990 I became a committee member. We held the initial CMT seminars at a community centre in Burwood as we tried to grow the support group and raise community and medical awareness of CMT. The opportunity came for us to convert an old storeroom at Concord Hospital thanks to Dr Nicholson and through the efforts of volunteers the CMTAA office was established. Our association began helping more people, providing support and information, directing those newly diagnosed to medical and allied professionals who had experience and knowledge in treating CMT and raising funds for research.

The CMTAA moved into larger premises at Concord Hospital; created a website and established support groups in state capitals and regional centres across Australia. In 2012, one of my twins Peter and his wife Jillian established CMT Aussie Kids. I hope people will come forward to keep the CMTAA going; they are needed. During my time as a volunteer, I felt that I was helping people and I know that all the volunteers feel a great level of personal satisfaction knowing we could talk to and understand the problems that people with CMT have to deal with.

I know that the CMTAA has helped raise awareness in the medical field and CMT is being diagnosed more now, it's no longer known as a rare disease. I have been asked to be a medical exam patient many times. When I first started none of the doctors could tell what I had; the usual response was that I had polio. The last time I attended all five trainee doctors diagnosed CMT. I felt like kissing them all.

Background photo: Phyllis's father Tom (right) was a fisherman on Botany Bay.

CMT expertise

Professors' Marina Kennerson & Steve Vucic

Australia continues to make significant contribution to Charcot-Marie-Tooth (CMT) research. Professor Kennerson, Director of the Northcott Neuroscience Laboratory at the ANZAC Research Institute, working in close collaboration with Professor Steve Vucic, Director of the Brain and Nerve Research Centre, Concord Hospital. Together they are pushing the frontiers of genetic discovery for unsolved CMT families and developing models for testing treatment therapies for these newly discovered genes.

Professor Vucic leads a hereditary nerve adult clinic at the Brain and Nerve Research Centre, which manages all inherited neuropathies including CMT, motor neuropathies, sensory neuropathies and inherited neuronopathies. As part of patient recruitment into the research program, his team of dedicated neurologists provide patients with careful phenotyping and electrophysiology studies as well as access to genetic testing through the Molecular Medicine Laboratory at Concord Hospital. Professor Vucic maintains a CMT patient registry which is critical for natural history studies and clinical trial readiness.

Professor Kennerson has worked for over 25 years in the field of CMT research and leads the CMT Gene Discovery and Translational Functional Genomics program at the ANZAC Research Institute. Marina has discovered numerous CMT genes, with her team conducting influential pioneering research to investigate structural variation in the non-coding or "dark genome" to identify new mutations in unsolved CMT families. Using cutting edge technologies, the novel mutations are investigated in patient nerves using induced pluripotent stem cells (iPSC) and modelled in living organisms (C. elegans) to develop biomarkers and gene therapy targets. Professor Kennerson is a board member of the Charcot-Marie-Tooth and Related Neuropathies Consortium (CMTR), Chair of the Asian Oceanic Inherited Neuropathy Consortium (AOINC) and serves on the Scientific Advisory Board of the CMT Research Foundation, USA.

The CMT research vision of Professor Kennerson and Professor Vucic is that discoveries from the Gene Discovery and Functional Genomics Program are translated into improved care and management for all CMT patients, as well as the ultimate development of therapies for clinical trials. The cornerstone of their research, and what makes this program possible, is the close collaboration between all stakeholders including, patients, carers, families, CMT Australia, researchers and clinicians. Continuing our close partnerships that nurture CMT research in Australia, will ensure successful productive outcomes as we move forward together.

Professor Garth Nicholson

Australia has a proud history of contributing to CMT research and Prof. Nicholson has been at the forefront of this research from the 'early days'. While conducting important research into CMT, Professor Nicholson realised the need for a member-based support network. Thus, in 1988 a fledgling Charcot-Marie-Tooth Association of Australia came into being. By investigating families, he mapped the genetic defects in the most common hereditary peripheral neuropathy (CMT1A) and familial forms of motor neurone disease (MND). Undoubtedly, Professor Garth Nicholson has made a significant global impact to research into CMT, and CMT Australia through his support and advocacy. We all owe a debt of gratitude to him.

CMT expertise

Professor Joshua Burns

Professor Joshua Burns, Ph.D., is Professor of Paediatric Neuromuscular Rehabilitation and Head of School and Dean of the Sydney School of Health Sciences in the Faculty of Medicine and Health at the University of Sydney Australia. He co-leads the Peripheral Neuropathy Management Clinic at the Children's Hospital at Westmead and is the Director of the Paediatric Gait Analysis Service of New South Wales at the Sydney Children's Hospitals Network.

He is a valued member of CMT Australia with a long list of publications and achievements in health science. His early research established risk factors for leg pain: an area he continues to research. His PhD thesis established assessment and treatment methods for foot deformity of adult neuromuscular disease. In 2009 Professor Burns completed an NHMRC Australian Clinical Research Postdoctoral Fellowship in the field of Neurology evaluating drug therapies for paediatric inherited neuropathies. An NHMRC Career Development Fellow (2011-2014), Professor Burns investigated treatments for childhood CMT and is particularly interested in developing and evaluating new treatments for children and adults with neurological and musculoskeletal disorders.

Dr Ché Fornusek

Diagnosed with CMT at the age of 6, later genetically diagnosed with CMT type X linked, Ché Fornusek was born and raised in New Zealand.

Ché has always adopted a positive approach to living with CMT. Dr Ché Fornusek, who lives in Sydney, is a senior lecturer and researcher in Exercise and Sport Science at the Faculty of Medicine and Health, University of Sydney. Drawing on his passion for sport and his interest in using exercise to promote health for people with a disability,

Ché had much to do with the establishment of Physical Disability Rugby League Australia and has represented New Zealand in 6 matches. Ché holds a biomedical engineering and physiology degree and completed his PhD in 2005.

Ché's research focuses on exploring the potential of sport, voluntary exercise, and electrical stimulation exercise to promote health in people with neurological injury or disease. His research examines the long-term benefits of exercise and physical activity for persons with neurological injury or disease. People with physical disability have higher rates of diabetes, cardiovascular disease, and obesity in comparison to the normal population.

The causes of these increased incidence rates are due to lower levels of activity, as well as greater barriers to exercise. Exercise or sport can be used to promote health for persons with disability.

Would you like to make a Donation?

CMT Australia is a registered charity, supported by dedicated volunteers throughout Australia. Every donation enables us to fund important research and subsidise running costs. All donations of $2 and over are tax deductible. To support this valuable cause, please fill out the online form https://www.cmt.org.au/control/donate or scan below.

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acnc.gov.au/charityregister

CMT AUSTRALIA
Charcot-Marie-Tooth Association Australia Inc.

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