



CMTAA
Charcot-Marie-Tooth Association Australia Inc.

**Moving
forward with
CMT**



In Association with
**THE UNIVERSITY OF
SYDNEY**

Welcome

The Charcot-Marie-Tooth Association of Australia Inc. (CMTAA) welcomes you to our community! Receiving a diagnosis of Charcot-Marie-Tooth (CMT) can be unsettling and overwhelming, but over the years we have acquired a vast array of knowledge about CMT and we are happy to share this with you. This booklet is an introductory guide; more specific information is available to members via our newsletter, national office, website (www.cmt.org.au) or by attending any of our seminars or support groups.



If you have recently been diagnosed with Charcot-Marie-Tooth Disease (CMT) you will find enormous benefit in building up a good health care team and a support team. An important part of understanding CMT is knowing other people with the disease, that is where the Charcot-Marie-Tooth Association of Australia Inc. (CMTAA) can help you and your family.

I was in my 40s when diagnosed with CMT. Having never heard of CMT I was at a loss to understand and get help. It was through the CMTAA that I found answers to my questions and, most importantly, a network of positive, approachable and enthusiastic people that allowed me to move forward.

Now, as President, I invite you to join the CMTAA, trusting that you can gain the same benefits as I have, including access to support groups and the latest research. Visit our website cmt.org.au where you will find information on becoming a member. I look forward to meeting you in the future.



Sue Hardy
President CMTAA

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Charcot-Marie-Tooth Association Australia Inc. ABN 63 076 189 912



CMTAA
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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. 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 also known by another name; Hereditary Motor and Sensory Neuropathy (HMSN). CMT is really an 'umbrella term' for a group of many hereditary diseases that affect peripheral nerves, hence the term, HMSN. Accordingly, CMT is divided into types and subtypes. Currently, over 80 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.

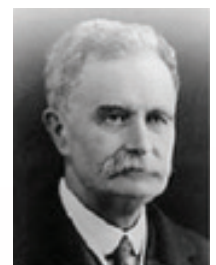
Although there is no cure for CMT, there are treatments that can be used to effectively manage its symptoms. Ongoing research includes efforts to slow down or even reverse nerve degeneration and muscle atrophy.



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.

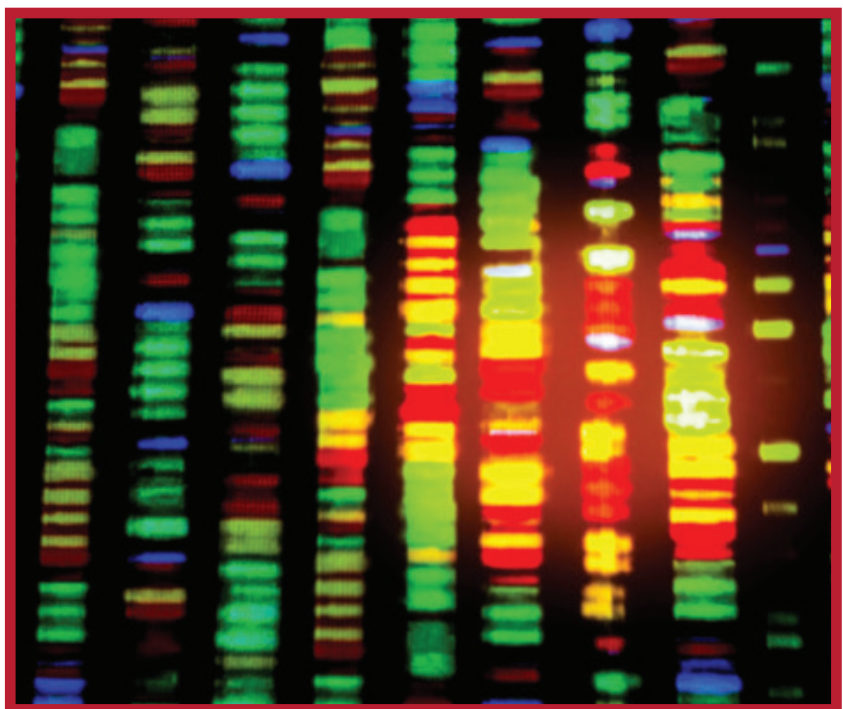
Formal Diagnosis

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 in vitro 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

It is to be expected you will need to take medications for conditions not related to your CMT. The CMTAA is supporting a study by Dr Alison Shield at the University of Canberra to capture adverse reaction experienced by people with CMT to medication(s). This study will improve the body of evidence on medication safety for people with CMT. The USA CMT Association provides a list of potentially harmful medication, but small numbers of adverse

reports in the CMT North American Database does not provide solid evidence. Consequently, the impact on people with CMT is unknown or unclear for most medications on this USA CMT Association 'potentially toxic' list. One exception is Vincristine, a cancer drug, where strong evidence exists for neurotoxicity.

If you are concerned how a medication might impact on you, free resources to assess medicine safety

are available from the National Prescribing Service Medicines Line 1300 MEDICINE or online at www.nps.org.au.

It is recommended to seek out health practitioners, be it your family doctor, neurologist and pharmacist who will answer questions and actively investigate medication safety 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 treatments 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 report this to the Adverse Medicines Event Line (1300 134 237) or online at the Therapeutic Goods Administration www.tga.gov.au. This can be done personally or in consultation with your health practitioner.

CMT

and your feet

Preventing and treating foot problems related to CMT

For people with CMT, correct footwear is critical, but it is difficult to find well fitting shoes due to high arched feet and hammered toes. Recent advancements in custom-made shoes and orthoses allow fashionable styles with innovative features designed to help anyone feel good on their feet and promote a healthy lifestyle.

People with CMT should view all foot difficulties as potentially dangerous and seek immediate podiatric care. Sensory loss, hypersensitivity and structural changes in the feet can result in pain as well as lessen the ability to feel pain, heat or cold. Decreased sensation may cause ulcers and infection to go unnoticed. Frequent self-inspection of your feet is essential. See your family doctor

or podiatrist rather than cut corns, calluses or ingrown nails yourself.

Reduced foot sensation requires special care against foreign objects when barefoot. Blisters, cuts and scrapes can be serious problems. Unattended ingrown nails can develop into infection. People with CMT are prone to fungal foot infections, which can make nails thick and painful. Preventative solutions such as correct socks avoid secondary bacterial infections that require antibiotics treatment.

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.

People affected by CMT often first display distinct physical symptoms that manifest in their feet.

- hammer toes
- very high arches
- poor sensation
- weak ankles
- foot drop

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



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.



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

1 High-arched feet (Pes cavus) is a very common characteristic. CMT causes weak foot muscles and in-turn the bones those muscles operate move and trigger bone and joint deformities. With reduced sensation, added pressure over protruding bones may not be felt, thus a danger of developing a sore, infection, or an ulceration.

2 Calluses often appear on the balls of feet due to pressure. Excess callus tissue may lead to ulcers. Ulcers may also form due to poor

circulation, inadequate soft tissue protection, infection, and pressure points caused by foot deformities and ill-fitting shoes. Most ulcers can be healed if treated early.

3 Ankle sprains are frequent for people with CMT. Deformities of ankles and feet due to progressive muscular weaknesses may cause imbalance of opposing muscle groups. Treatment may range from bracing with orthoses, deep shoes with soft inlays or surgical realignment. Maintaining flexibility and strength is



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

recommended with regular stretching and exercise. Custom-moulded braces and in-shoe orthoses can greatly assist people with CMT. In recent years, new foot surgery techniques to realign the ankle joint have shown some success.

4 Hammer toes (clawed toes) and corns on the toes are usual. The muscles within the foot itself (intrinsic muscles) weaken, and cause the toes to curl up. Various treatments include toe splints, insoles and surgery to straighten toes.

5 Weakness in calf muscles often cause foot drop. To reduce the instances of tripping due to foot drop, carbon fiber lightweight ankle-foot orthoses (AFO's) that fit into most shoes and are easy to fit around the calf show considerable success. While effective treatments include AFO's and rocker bottom-style shoes, in some instances surgical transfer of stronger muscles to the top of the foot can also help stabilize foot drop.

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.”



CMTAA youth camps provide participants with a holiday setting that includes activities such as kayaking, archery, abseiling as well as networking with other participants. Analysis of participant testimonial letters and numbers of anecdotes show positive experiences following these camps.

Most children had not met someone with CMT of their age before. Those that attended camps reported that they had felt isolated, that there was no awareness surrounding their illness, and that they were glad to be in touch with other children sharing their journey.

The CMTAA offers support for children with CMT. A youth camp is held annually in order for children to meet with peers; share experiences, challenges and successes; explore and enjoy challenging and adventurous activities in a safe environment; and to learn more about CMT.



Aussie Kids Camp adult leaders are all dedicated volunteers who either have CMT themselves, or are closely related to someone with CMT. Volunteers have experience in education, law enforcement and youth organisations. All undergo Working with Children checks.

A recurring positive theme is found in testimonials; 'uplifting, confidence, interaction and belonging.' Numerous studies have been conducted on the importance of support groups; with reward ranging from empowerment and knowledge-sharing to psychological benefits.

CMT Aussie Kids National Camp

CMT Aussie Kids, an activity of CMTAA, runs a national camp exclusively for youth living with CMT. CMT Aussie Kids Camp is open to youth aged from 8 to 18 from across Australia, where 'kids' can meet with their peers to share their experiences, challenges and successes; explore and enjoy challenging and adventurous activities in a safe environment; and learn about managing CMT.

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

In addition to the camp, a 'CMT Aussie Kids' Facebook page allows youth living with CMT to connect and support each other through social media. Strictly youth focussed, adult membership is restricted. Members of this closed Facebook group do not need to have been to one of the camps but should be prepared to participate and support their peers.

As the camp evolves, and the kids grow, we encourage young adults as 'youth leaders' and role models to the newer attendees, further developing a sense of achievement, responsibility as well as positioning our CMT youth for future roles in the community.

Interested? For further information contact the CMTAA or email cmtaussiekids@gmail.com.

CMTAA

Benefits of membership

The CMT Association Australia Inc. (CMTAA) began as an informal support group in 1988, and was incorporated in 1990 as a registered charity. The motivation to establish the CMTAA was to meet the need for people with CMT to communicate with each other and provide mutual encouragement. While mutual support is still a core aim of the CMTAA, its 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 and social media

- 5** Promote CMT awareness to medical and allied 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 the CMTAA to join our management team.



"After attending just one CMTAA 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 speaking to findings from his Department of Health study 'Reducing the Health Burden of CMT in Australia' for the 2015 CMTAA Annual Seminar at Concord Repatriation Hospital.

CMT Community



The Critchley family

The Critchley family; Jillian, Peter, Matilda and Eleanor are often seen as the 'face' of CMT in Australia having appeared on ABC's science program Catalyst, SBS' Insight, CMTAA seminars as well as print and social media. Peter and his two daughters Matilda and Eleanor have CMT type 1A.

Both Jillian and Peter are active members of the CMTAA, participating in the National Committee and annual seminars. Jillian is editor of the CMT CoMmuniTy newsletter for CMTAA members. In 2012

Jillian and Peter founded 'CMT Aussie Kids', aimed at linking youth living with CMT through an annual national camp. Jillian and Matilda moderate a 'CMT Aussie Kids' Facebook page which connects and offers support to young people living with CMT.

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 entered the nursing profession.

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. Matilda and Eleanor are an integral part of the development of the CMT Aussie Kids program, providing support, encouragement and understanding, particularly to new members who may feel anxious meeting others with the same condition.

Isabella Frisan

Diagnosed with CMT at 7, Isabella has not allowed her condition to define her, meeting every challenge with a positive approach.

Isabella's choice to pursue her love for creativity shows in her ability as an accomplished musician (majoring in piano and percussion). Since the age of 8, Isabella has participated in solo, ensemble, orchestral, theatrical and dance performances, achieving many awards in music competitions. Isabella has found that her passion for the Arts has enabled her to give back to the community through volunteer work.

Dr Grace Warren's 'use it or lose it' message has made a big influence on Isabella. She believes playing the piano has improved coordination, flexibility and strength in her hands.

At age 10, Isabella was scouted for the Australian Paralympic swimming training program.

At 19, Isabella moved from her home in Adelaide to Canberra to undertake Law and Commerce (Hon) at the Australian National University. Isabella aspires to be more involved in disability advocacy. Her drive is an inspiration to all, making Isabella a valued CMTAA team member.



CMT Community



Peter Richards

Peter Richards and his wife Liz have lived in Catherine Field NSW for over 40 years where they raised two boys. Diagnosed with CMT, there is not much Peter hasn't done. Soccer coach for his son's team, climbing the Harbour Bridge, skydiver, golfer, skier, cyclist and commercial pilot; Peter's passion for life is inspirational. At 70 years of age, Peter still maintains his lifetime passion for breeding and training horses. Always awake at the crack of dawn and a regular at the local gym, Peter and Liz continue to run their own publishing company.



Mason Anderson

Mason Anderson from Mildura Victoria was diagnosed with CMT when he was 14 years old. After high school, he worked for a year at a local Mildura law firm. Now 19 years of age, Mason has moved from his family home in Mildura to Adelaide where he is a full-time law student. On top of study, Mason manages to hold down a part-time job in security. He also enjoys training horses and regularly works out at the gym. Mason has a well-rounded proactive approach to manage his CMT.



Roger MacRury

Not only can Roger MacRury fly a plane, he can build them. Roger lives in Canberra and hangers the plane he co-built in Goulburn. A very well respected member of the CMTAA, Roger enjoys that flying gets him places, including flying to visit family in Melbourne. Roger was not formally diagnosed until he was 45 although the symptoms were evident from early childhood. Now 63 and retired, Roger has enjoyed a long career in the public service for the Bureau of Meteorology, Treasury, Health and Defence.

CMT expertise

About Dr Ché Fornusek

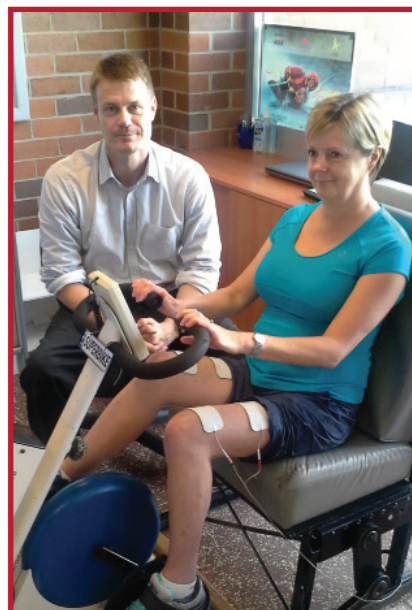


Diagnosed with CMT at the age of 6, later genetically diagnosed with CMT type X linked, Ché Fornusek was born and raised in New Zealand. Ché has always adopted a positive approach to living with CMT. Dr Ché Fornusek, now 44, married and living in Sydney, is a lecturer and researcher in Exercise and Sport Science at the University of Sydney, Faculty

of Health Sciences. 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 NSW Physical Disability Rugby League. Ché holds a biomedical engineering and physiology degree and completed his PhD in 2005. Ché has taught across the full range of programs in the Faculty of Health Sciences, from first year undergraduate to postgrad coursework and PhD research assistant supervision, as well as rehabilitation engineering courses. 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.

Ché's 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 compared to the normal population. The cause of the increased incidence rates are due to lower levels of activity but also greater barriers to exercise. Exercise or sport can be used to promote health in persons with disability.



If you could give your 18-year-old self advice, what would it be?

When I was young I didn't want to show that I had a disability and I felt as though it was really important to be like everyone else, and not to stand out. Working in a competitive environment (including as an academic at the University) you need to stand out; so changing this mind-set has been difficult for me. I would tell myself to embrace my disability, to an extent, and to not worry too much about being different in that way. It becomes an advantage in the future: it can change your behaviour and understanding of other people, set your thinking differently, and inspire you to learn things quickly.

CMT

expertise

About Professor Garth Nicholson

Australia has a proud history of contribution to CMT research. Professor Garth Nicholson at the University of Sydney initiated the first search for the genes that cause CMT neuropathy, in a genome search using RFLP mapping probes. In 1989, he found the locus of CMT type 1A on the proximal short arm of chromosome 17. Professor Nicholson showed that the PMP22 gene was included in a large DNA duplication which resulted from misalignment between two large flanking repeat regions.

This innovative use of molecular methods to locate gene mutations causing peripheral nerve diseases stimulated interest in the area. These discoveries facilitated development of the first CMT

support organization (the CMT Association of Australia) and commencement of a CMT clinic and country outreach workshops. A pes cavus (high foot arch) prevention program was developed with physiotherapists and orthopaedic surgeons. These advances were later adopted in London, Oxford, USA and Children's Hospitals in Australia.

Professor Nicholson has over 150 refereed journal publications and five papers in Nature Genetics, two papers in Science and over 3000 citations. Garth has contributed to and has chaired international scientific meetings and is on the editorial boards of the journals Muscle and Nerve and Amyotrophic Lateral Sclerosis and is a committee



member of the Charcot-Marie-Tooth Association of the USA. He is Patron of the Machado-Joseph Disease Foundation and is on the Neurogenetics committees of the Neurological Associations of Australasia and the USA. Garth serves on the External Advisory Board of the Inherited Neuropathy Consortium of the USA.



About Professor Joshua Burns

Professor Joshua Burns combines research and clinical training in Podiatry, Physiotherapy and Neurology. He is a valued member of the CMTAA 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. He is Director of the Paediatric Gait Analysis Service of New South Wales at the Sydney Children's Hospital's Network (Randwick and Westmead) and Co-Director of the Arthritis and

Musculoskeletal Research Group at The University of Sydney. 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.

The Douglas Legacy A hereditary journey

Phyllis Critchley (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 and his wife 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.

Would you like to make a Donation?

Being a Charity, every donation is highly valued and enables us to further our research and subsidises running costs for this valuable cause.

All donations of \$2 and over are tax deductible.

If you would like to support us please fill out the online form <https://www.cmt.org.au/control/donate>



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