REDUCING THE HEALTH BURDEN OF CHARCOT-MARIE-TOOTH IN AUSTRALIA 2015

The Chronic Disease Prevention and Service Improvement Fund is an Australian government initiative administered by the Department of Health and Ageing.
Acknowledgement

This report ‘Reducing the health burden of Charcot-Marie-Tooth disease in Australia’ is dedicated to the hard working members of the CMT Association of Australia Inc. (CMTAA) who give their time, passion and tireless enthusiastic support free of any restitution.

The 418 response survey (Charcot-Marie-Tooth in Australia: A Survey of Cost-Utility Wellbeing and Management) together with 21 focus groups comprising 178 participants held every state of Australia and the ACT would not have been possible without the huge contribution of the CMTAA Executive Committee. Our gratitude to all of you for your help is immense.

We are extremely grateful to the CMTAA regional organisers who offered their total commitment in partnership with the author to organise and host focus groups Australia wide. Special thanks to Anycie Berkmann who, was instrumental in the distribution of the hardcopy and online surveys. Special thanks also to CMTAA Treasurer Robert Twin for overseeing the financial elements crucial to the project’s success.

Lastly, one person warrants very special mention above all others for his unwavering commitment to seeing this project to completion. CMTAA President Darryl Beitsch who, from the genesis of this ambitious undertaking was responsible for the organisational aspects of the survey, from encouraging on the members to mailing the forms.

Darryl, we owe you a great debt of thanks.

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2015
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Abbreviations

ABS-Australian Bureau of Statistics
ACT-Australian Capital Territory
ASGS-Australian Statistical Geography Standard
BoD-Burden of Disease
CGH-Comparative Genomic Hybridisation
CMT-Charcot-Marie-Tooth
CMTA-Charcot-Marie-Tooth America
CMTAA-Charcot-Marie-Tooth Association of Australia Incorporated
DDA-The Disability Discrimination Act 1992
DSP-Disability Support Pension
DSS-Dejerine-Sottas syndrome
GDD-Global Development Delay
HREC-Human Research Ethics Committee
IVF-In vitro fertilization
NCV-Nerve Conduction Velocity test
NDIS-National Disability Insurance Scheme
NPS-Neuropathic Pain Scale
NT-Northern Territory
OSA-Obstructive sleep apnoea
PGD-Preimplantation genetic diagnosis
PIS-Participant Information Sheet
PwCMT-Person with Charcot-Marie-Tooth
Qol-Quality of Life
RA-Regional Area
RLS-Restless legs syndrome
SDAC-Survey of Disability, Ageing and Carers
SCWM-CMT in Australia: A Survey of Cost-Utility, Wellbeing and Management
TTSS-Taxi Transport Subsidy Scheme
USYD-The University of Sydney
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Definitions

Core activity limitation
Four levels of core activity limitation are determined based on whether a person needs help, has difficulty, or uses aids or equipment with any of the core activities (mobility or self care).

A person's overall level of core activity limitation is determined by their highest level of limitation in these activities. The four levels of limitation are:

**profound**: the person is unable to do, or always needs help with, a core activity task.

**severe**: the person sometimes needs help with a core activity task

**moderate**: the person needs no help but has difficulty with a core activity task

**mild**: the person needs no help and has no difficulty with any of the core activity tasks, but uses aids and equipment

- cannot easily walk 200 metres
- cannot walk up and down stairs without a handrail
- cannot easily bend to pick up an object from the floor
- cannot use public transport
- can use public transport but needs help or supervision, or needs no help or supervision but has difficulty using public transport.

Mobility
Mobility comprises the following tasks:

- getting into or out of a bed or chair
- moving about the usual place of residence
- going to or getting around a place away from the usual place of residence
- walking 200 metres
- walking up and down stairs without a handrail
- bending and picking up an object from the floor using public transport.

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Schooling restriction
A schooling restriction is determined for persons aged 5-20 years who have one or more disabilities if, because of their disability, they:
- are unable to attend school
- attend a special school
- attend special classes at an ordinary school
- need at least one day a week off school on average, or have difficulty at school.3

Self care
This activity comprises the following tasks: showering or bathing, dressing, eating, toileting, and bladder or bowel control.

Long-term condition
A disease or disorder which has lasted or is likely to last for at least six months; or a disease, disorder or event (e.g. stroke, poisoning, accident etc.) which produces an impairment or restriction which has lasted or is likely to last for at least six months. Long-term health conditions have been coded to a classification based on the World Health Organisation's International Classification of Diseases, 10th Revision (ICD-10).

Disability
Disability is defined using the International Classification of Functioning, Disability and Health (ICF),4 which, in the context of health experience, defines disability as an umbrella term for impairments, activity limitations and participation restrictions. It denotes the negative aspects of the interaction between an individual (with a health condition) and that individual’s contextual factors (environment and personal factors).

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The SF-36 scale

The SF-36 can evaluate the impact of a disorder such as CMT on eight aspects of a person’s life. The eight aspects (or dimensions) are:

- Physical function – the person’s physical ability to do things.
- Role Physical – the impact of any physical limitation on the person’s life and relationships.
- General Health – an overall measure of the physical health of a person.
- Vitality – a measure of one’s feeling of well being, or of ‘joie de vivre’.
- Social Function – the impact of a disorder on the more behavioural aspects of social function.
- Emotional Role – the impact of a disorder on the person’s ability to fulfil the emotional aspects of their life and relationships.
- Mental Health – the degree to which the disorder leads to feelings of anxiety or depression.

The SF-36 scores in each dimension range from 0 to 100, with 100 theoretically perfect and 0 as bad as possible, (higher scores = better health).\(^5\)

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DESIGN AND METHOD Study Population

Primary data was collected in two formats: (1) A survey for which the criterion was that the participant be over 18 years of age, living in Australia and diagnosed with CMT, and (2) Focus groups for which the criterion was that the participant have CMT, be over 18 years of age, living in Australia, a family member, carer or partner of a pwCMT.

The Survey

a. Survey Construction

The survey ‘Charcot-Marie-Tooth in Australia, a survey of cost-utility, well-being and management’ was constructed in hard copy and electronic format (for personal computer or smart phone). The survey was designed to examine economic cost, QoL and key elements of living with CMT (see Table 1). The Project Manager Dr Scott Denton drew on his expertise in survey design with the Australian Bureau of Statistics as well as his own experience as a pwCMT (Dejerine Sottas). Respondent effort was a concern because of the survey’s length (200 questions over 16 pages), with emphasis on visual layout, question wording and sequence in a multiple-choice format. To further stimulate respondent frequency, the survey was designed to be completed in one sitting. Survey completion in time trials averaged 34 minutes for the hardcopy version and 32 minutes for the online version.

b. Survey Participation

Survey participation was limited to adult Australians with CMT. To avoid duplication, each survey had an individual code number; however no data was linked to any individual participant in accordance with USYD HREC requirements. Primary recruitment in the study was through the CMTAA, which is the sole CMT support and advocacy organization in Australia, with regional coordinators holding regular meetings of support groups in every Australian state and territory. The CMTAA began as a support group in 1988, and is now a member-based registered charity, which advocates and offers support and management to improve the lives and conditions of people affected by CMT. After a solid marketing approach in the CMTAA’s quarterly newsletter, 490 CMTAA financial and non-financial members post-2012 were invited to participate in the study. Secondary recruitment was done through encouraging CMTAA
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members to invite non-CMTAA family members to participate, given that a single CMTAA member might have family members with CMT who are not CMTAA members.

c. Survey Responses

At 418 useable responses, the survey is the largest of its kind to date. The hardcopy version returned 260 and the online version returned 158 responses. Given a primary recruitment of 716, the high frequency of response should be noted, principally in consideration of the large size of the survey. Participants ranged from 18 to 96 years of age, and were drawn from every state and territory. The first conclusion of this study is that pwCMT are a motivated group willing to assist researchers and have their voice heard. Preliminary SCWM data analysis often identified areas of interest to then shape focus group questions.

<table>
<thead>
<tr>
<th>Survey out</th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Hardcopy</td>
<td>CMTAA members</td>
<td>254</td>
</tr>
<tr>
<td>Softcopy</td>
<td>CMTAA members</td>
<td>234</td>
</tr>
<tr>
<td>Hardcopy</td>
<td>Non-members</td>
<td>197</td>
</tr>
<tr>
<td>Softcopy</td>
<td>Non-members</td>
<td>31</td>
</tr>
<tr>
<td>Total</td>
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<td>716</td>
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</table>

<table>
<thead>
<tr>
<th>Survey in</th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Hardcopy</td>
<td>CMTAA members</td>
<td>207</td>
</tr>
<tr>
<td>Softcopy</td>
<td>CMTAA members</td>
<td>134</td>
</tr>
<tr>
<td>Hardcopy</td>
<td>Non-members</td>
<td>53</td>
</tr>
<tr>
<td>Softcopy</td>
<td>Non-members</td>
<td>24</td>
</tr>
<tr>
<td>Total</td>
<td></td>
<td>418</td>
</tr>
</tbody>
</table>
The Focus Groups

a. Focus group overviews

The second data collection phase involved qualitative interviews through focus groups. Twenty-one groups were organised, in every state of Australia and in the ACT. A total of 178 people were interviewed, comprising pwCMT and their families. Each group lasted 60 minutes and was audio-taped, with participants asked open-ended questions designed to stimulate discussion. Participants were provided with a PIS and consent form. An Interpreter assisted with one focus group of non-English speaking pwCMT of Vietnamese heritage. Focus group audiotapes were destroyed after transcription, and identifying information was removed in compliance with USYD HREC requirements.

b. Focus group questions

The questions were presented in the form of conversations and the proceedings were deliberately kept fluid. Focus group questions followed the same theme and structure as that of the survey, with 13 topics in all: (1) demographic data (i.e. age, height, weight and sex); (2) lifestyle and life satisfaction; (3) living situation; (4) relationships; (5) health care and individual health (i.e. CMT type, children diagnosis, pain and pain management); (6) medication; (7) mobility and exercise; (8) sleep and fatigue; (9) medical (i.e. surgical, genetic counselling); (10) diet and nutrition; (11) alcohol and tobacco; (12) education; (13) employment and income.

c. Analysis of the Interviews

Transcriptions were organized into themes that identified and described the experiences of living with CMT, and life issues surrounding people with CMT. Comments and narratives from participants were then grouped under each theme. Detailed categorization of the comments and narratives within the themes was then identified, with the aim of highlighting the impact of the specific diagnosis of CMT on an individual.

d. Focus group and survey union

The combination of qualitative and quantitative techniques of focus groups and survey into a single research design proved to be a highly successful methodological union. Focus groups offered flexible qualitative approaches to peruse particular issues in further depth as well as accommodating a wider range of explanatory categories unforeseen in the original survey.
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design. Focus groups allowed an important survey gap to be filled, that of children with CMT. While survey participation was restricted to adults, participants who were parents were asked ‘benchmark’ questions (i.e. child’s sex, child with CMT Y/N, age, living at home Y/N, etc.). The qualitative depth of focus group data further complemented the quantitative survey approach in additional question themes (i.e. relationships, mobility, pain recognition and pain management).

e. Map of Australia showing focus groups locations
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1. Introduction

What is Charcot-Marie-Tooth (CMT)?

Charcot-Marie-Tooth (CMT) is the name given to a genetically and clinically heterogeneous group of inherited disorders of the peripheral nervous system, first described in 1886 by Jean-Martin Charcot, his pupil Pierre Marie, and Howard Henry Tooth. CMT is the most common type of inherited neurologic disorder. It is estimated that it affects 1 in every 2,500 people. This equates to approximately 9,552 people in Australia, although this figure is conservative, given probable rates of under-diagnosis. CMT affects the peripheral nerves — those that leave the spinal cord and travel to the feet and hands. It is a length-dependent condition that affects the nerves at the farthest points first and then progresses proximally. CMT affects both motor and sensory nerves. It is characterised by progressive loss of muscle tissue and touch sensation, and can cause foot drop and walking difficulties, as well as problems with balance and neuropathic pain. Other signs and symptoms can occur depending on the type of CMT.

Severity of clinical signs accompanying CMT is highly variable, even within families with the same CMT type. Further, the impact on pwCMT can range from the sub-clinical, to significant disability necessitating mobility aids from braces and splints up to wheelchairs. Most newly diagnosed, for themselves or their children, pursue knowledge about the prevalence of the various secondary features and the possible future impact of the disease. Regrettably, practically all of the existing material is anecdotal. Suitable information is limited although neurologists understand secondary features. The foremost obstacle to the gathering of data on secondary features is the comparatively low frequency of CMT in the Australian population. Low frequency pooled with the variety of secondary presentations creates further difficulties. Consequently, it has proved problematic previously to assemble a large enough sample of pwCMT to collate a comprehensive spectrum of epidemiological data. To survey CMTAA members and include validated instruments was an invaluable opportunity.

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Program Description and Objectives
The Chronic Disease Prevention and Service Improvement Fund (the fund) is an Australian government initiative administered by the Department of Health and Ageing (the Department) designed to support activities that address the rising burden of chronic disease. The objective of the fund is to support targeted action related to chronic disease prevention and service improvements, particularly within the primary care and community sectors.

The fund aims to:

Reduce the incidence of preventable mortality and morbidity;

• Maximise the wellbeing and quality of life (QoL) of individuals affected by chronic disease from initial diagnosis to end of life;
• Reduce the pressure on the health and hospital system including aged care; and
• Support evidence-based best practice in the prevention, detection, treatment and management of chronic disease.

Aims
The aims of this project were to:

• Maximise the wellbeing and quality of life of individuals affected by CMT throughout their whole life span.
• Ascertain what the cost of CMT is to government and recommend policies and strategies to better utilise resources within the identified sections of the public sector.
• Establish evidence-based best practice to support the prevention and management of CMT.

Priority areas
The following priority areas have been identified to achieve the Fund’s objectives and translate its key principles into practice:

• Prevention across the continuum;
• Early detection and appropriate treatment;
• Integration and continuity of prevention and care; and
• Self-management.
Types of CMT

There are many genetic variations of CMT. Known subtypes are listed in the CMT subtypes table below. Estimates vary as to the frequency of CMT subtypes.\(^8\) CMT1 accounts for about 50% of CMT, while CMT2 and CMTX account respectively for 10% to 15%. DI-CMT forms are rare. CMT1A accounts for 70–80% of CMT1, CMT1B for about 10%. CMT2A2 accounts for about 20% of CMT2. CMTX1 accounts for 90% of all CMTX.\(^9\)

### CMT subtypes table\(^10\)

<table>
<thead>
<tr>
<th>Subtype</th>
<th>Gene</th>
<th>Chromosome</th>
<th>Distinctive features</th>
</tr>
</thead>
<tbody>
<tr>
<td>CMT1A</td>
<td>PMP22</td>
<td>17p12</td>
<td>Most common subtype, onion bulbs nerve biopsy</td>
</tr>
<tr>
<td>CMT1B</td>
<td>MPZ</td>
<td>1q23.3</td>
<td>Often identical to CMT1A, late-onset axonal variant</td>
</tr>
<tr>
<td>CMT1C</td>
<td>LITAF/SIMPLE</td>
<td>16p13.13</td>
<td>No distinctive features</td>
</tr>
<tr>
<td>CMT1D</td>
<td>EGR2</td>
<td>10q21.3</td>
<td>Rare subtype, no distinctive features</td>
</tr>
<tr>
<td>CMT1E</td>
<td>PMP22, MPZ, Cx32, others</td>
<td></td>
<td>Association of CMT with deafness</td>
</tr>
<tr>
<td>CMT1F</td>
<td>NEFL</td>
<td>NEFL</td>
<td>mutations also associated with CMT2E</td>
</tr>
<tr>
<td>HNPP</td>
<td>PMP22</td>
<td>17p12</td>
<td>Asymmetric recurrent neuropathy, tomacula</td>
</tr>
<tr>
<td>HNA</td>
<td>SEPT9</td>
<td>17q25</td>
<td>Recurrent painful brachial plexopathies, dysmorphic features</td>
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<tr>
<td>CMTX1</td>
<td>Cx32</td>
<td>Xq13.1</td>
<td>Common subtype, CNS involvement, variable NCVs</td>
</tr>
<tr>
<td>CMTX2</td>
<td>Unknown</td>
<td>Xp22.2</td>
<td>Infantile onset, mental retardation</td>
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<tr>
<td>CMTX3</td>
<td>Unknown</td>
<td>Xq26</td>
<td>Prominent paresthetic pain, women with high arches</td>
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<td>CMTX4</td>
<td>Unknown</td>
<td>Xq24-q26.1</td>
<td>Severe, deafness and mental retardation in males</td>
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<td>CMTX5</td>
<td>PRPS1</td>
<td>Xq22.3</td>
<td>Axonal neuropathy, deafness, optic neuropathy</td>
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<td>CMTX6</td>
<td>PDK2</td>
<td>Xp22.11</td>
<td>Axonal polyneuropathy</td>
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<td>DI-CMTA</td>
<td>Unknown</td>
<td>10q24.1-q25.1</td>
<td>Second decade onset</td>
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<tr>
<td>DI-CMTB</td>
<td>DNM2</td>
<td>19p13.2</td>
<td>Mild–moderate, neutropenia, cataract, onion bulbs</td>
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<tr>
<td>DI-CMTC</td>
<td>YARS</td>
<td>1p35.1</td>
<td>Variable onset</td>
</tr>
<tr>
<td>DI-CMTD</td>
<td>MPZ</td>
<td>1q23.3</td>
<td>Variable distal wasting, weakness and sensory loss</td>
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<tr>
<td>DI-CMTE</td>
<td>INF2</td>
<td>14q32.33</td>
<td>Second decade onset, legs &gt; arms, kidney disease, hearing loss</td>
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<tr>
<td>CMT2A1</td>
<td>KIF1B</td>
<td>1p36.2</td>
<td>Rare</td>
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<tr>
<td>CMT2A2</td>
<td>MFN2</td>
<td>1p36.2</td>
<td>Common subtype, variable onset and severity</td>
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<tr>
<td>CMT2B</td>
<td>RAB7</td>
<td>3q21.3</td>
<td>Most sensory, foot ulcerations, occasional onion bulbs</td>
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<tr>
<td>CMT2B1</td>
<td>LMNA</td>
<td>1q22</td>
<td>Second decade onset</td>
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10 Ibid.
<table>
<thead>
<tr>
<th>CMT2B2</th>
<th>MED25</th>
<th>19q13.33</th>
<th>Minimally reduced NCVs</th>
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<tr>
<td>CMT2C</td>
<td>TRPV4</td>
<td>12q24.11</td>
<td>First decade onset, vocal fold dysfunction</td>
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<tr>
<td>CMT2D</td>
<td>GARS</td>
<td>7p14.3</td>
<td>Worse hand than leg weakness, slow progression</td>
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<tr>
<td>CMT2E</td>
<td>NEFL</td>
<td>8p21.2</td>
<td>Variable onset age and severity</td>
</tr>
<tr>
<td>CMT2F</td>
<td>HSPB1</td>
<td>7q11.23</td>
<td>Slow progression</td>
</tr>
<tr>
<td>CMT2G</td>
<td>Unknown</td>
<td>12q12-q13.3</td>
<td>Slow progression of foot deformity, conduction velocities may be normal</td>
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<td>CMT2H/K</td>
<td>GDAP1</td>
<td>8q21.11</td>
<td>Variable onset and severity, some with pyramidal signs</td>
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<tr>
<td>CMT2I</td>
<td>MPZ</td>
<td>1q23.3</td>
<td>Asymptomatic or cramps with foot deformities</td>
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<tr>
<td>CMT2J</td>
<td>MPZ</td>
<td>1q23.3</td>
<td>Fourth to sixth decade onset, deafness, sensory and autonomic dysfunction</td>
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<tr>
<td>CMT2L</td>
<td>HSPB8</td>
<td>12q24.23</td>
<td>Also linked to dHMN2A</td>
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<td>CMT2M</td>
<td>DNM2</td>
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<td>Prominent gait ataxia</td>
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<td>CMT2N</td>
<td>AARS</td>
<td>16q22.1</td>
<td>Onset first to sixth decade, asymptomatic or distal weakness/sensory loss/deafness</td>
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<td>CMT2O</td>
<td>DYN1C1H1</td>
<td>14q32.31</td>
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<td>DSS</td>
<td>PMP22, EGR2, MPZ, PRX</td>
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<td>Onset in infancy or early childhood, hypertrophic nerves</td>
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<td>GDAP1</td>
<td>8q21.11</td>
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<tr>
<td>CMT4B1</td>
<td>MTMR2</td>
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<tr>
<td>CMT4B2</td>
<td>SBF2</td>
<td>11p15.4</td>
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<td>CMT4C</td>
<td>SH3TC2</td>
<td>5q32</td>
<td>Prominent scoliosis; early loss of ambulation; heterozygotes: mildly affected</td>
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<td>Cranial nerve involvement, arthrogryposis, and respiratory failure</td>
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<td>PRX</td>
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<tr>
<td>CMT4G Russe</td>
<td>HK1</td>
<td>10q22.1</td>
<td>Severe disability, prominent sensory loss, intermediate motor NCVs</td>
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<td>12p11.21</td>
<td>Early onset, scoliosis, hypomyelination, redundant myelin sheaths</td>
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<td>CMT4J</td>
<td>FIG 4</td>
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<td>Severe phenotype, childhood onset, motor dominant</td>
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<td>CMT RIA</td>
<td>GDAP1</td>
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<td>Variable severity</td>
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<td>Self-abusive behavior and dysmorphic features</td>
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<td>GAN</td>
<td>Gigaxonin</td>
<td>16q23.2</td>
<td>Early onset, mental retardation, tightly curled hair</td>
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</table>
Age of onset

The onset of symptoms for the most common form of CMT, CMT1A, is usually in the first or second decade of life, although mild cases may become apparent only much later in life. Other rarer subtypes of CMT can be more severe, presenting in infancy or early childhood and progressing more quickly. As the disease usually advances slowly, children are often initially described as clumsy. Foot deformities, such as hammertoes, pes cavus (high arches) and pes planus (flat feet), are very common. Ninety-five percent of people with CMT1A are never wheelchair bound.\(^\text{11}\) Throughout this report, the term ‘late-onset’ denotes disease activity occurring over the age of 10, and ‘early-onset’ denotes disease activity under the age of 10.\(^\text{12}\)

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2. Diagnosis

CMT neuropathies comprise a group of monogenic disorders affecting the peripheral nervous system. More than one thousand distinctive mutations have been discovered in eighty disease-associated genes. The reference genome that resulted from the Human Genome Project together with progress in sequencing technologies has greatly enhanced gene and mutation breakthroughs.\(^\text{13}\)

"In fact, the first clinical whole genome sequence was reported in a patient with CMT."\(^\text{14}\)

Diagnosis is a particularly significant part of any individual’s CMT journey. Focus group data revealed that most people knew there was something wrong with their bodies well before they were formally diagnosed with CMT. In this pre-diagnosis period, for some spanning decades, pwCMT can face a range of emotional and physical challenges. Early diagnosis will lead to positive benefits in treating and managing CMT, as well as assisting with associated mental health issues such as anxiety and depression, thus may improve the overall QoL.\(^\text{15}\)

**Initial diagnosis**

Initially for many people, it is often a family member or a friend who is the first to notice the symptoms of CMT, which may include clumsiness, lower leg weakness or foot deformities (abnormally high arches, clawed or hammered toes).\(^\text{16}\)

"My daughter was walking up the stairs to accept an award on stage at school. Her movements were just not right. It was then I suspected something was wrong."

PwCMT may exhibit ‘champagne legs’, that is, where the leg is extremely narrow at the ankle and just above, but then widens in the upper part of the thigh above the knee, so as to give the appearance of an inverted champagne bottle.\(^\text{17}\) Other typical features include weakness of the foot, which may result in foot drop and a high stepped gait. Studies show that pwCMT often


\(^\text{14}\) ibid.


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trip (96.1%), have difficulty running (96.1%) or with slippery surfaces (95.8%), and fall over regularly.\textsuperscript{18} For many, such incidents were the first signs that something is wrong.

\begin{figure}[h]
\centering
\includegraphics[width=\textwidth]{who_diagnosed_cmt.png}
\caption{Who diagnosed your CMT?}
\end{figure}

\textbf{Formal Diagnosis}

SCWM data reveals that 77.3\% of pwCMT were first informed that they had CMT by a neurologist, with 5.7\% informed by their GP. A further 7.0\% were informed by somebody else, of which podiatrists (28\%) and physiotherapists (23\%) the majority, the remainder comprising of a diverse group including chiropractors and Dr Google.

Because of the hereditary nature of the disease, it is on occasion another family member (usually a parent) who informs the individual that they may have the disease (5.7\%). CMT may run in a family even without any noticeable family history. In part, this is because CMT can be inherited in three different ways that are difficult to trace through a family tree: X-linked, autosomal dominant and autosomal recessive.\textsuperscript{19} However the absence of a family history of CMT does not exclude the possibility of having the disease. Formal testing may be electro-diagnostic and/or genetic. Electro-diagnostic testing is commonly used to diagnose CMT through nerve conduction velocity testing (NCV), which calculates the strength and speed of

\begin{itemize}
\end{itemize}
electrical signals travelling along the peripheral nerves.\textsuperscript{20} Electro-diagnostic tests are typically performed by a neurologist. Genetic tests are available to examination for many but not all common chromosomal defects triggering CMT. A positive genetic test will provide conclusive diagnosis and can also contribute to useful knowledge for possible family planning. A negative result does not exclude CMT, given that certain types cannot be tested using DNA sampling.\textsuperscript{21}

\begin{figure}[h]
\centering
\includegraphics[width=\textwidth]{Age_of_Diagnosis.png}
\caption{Age of Diagnosis}
\end{figure}

Note: SCWM data age of diagnosis to frequency (total 401 respondents).

Across Australia, female and male groups and all CMT types, the average age of diagnosis for a pwCMT is 33 years of age, with a standard deviation of 18.22. The median age of diagnosis is 33 years and 6 months, with the most common age of diagnosis being 40. The highest frequency (12.2\%) of SCWM participant diagnoses fell within the age cohort of 45-49. The age of diagnosis covers the full spectrum from birth to 78 years. PwCMT can live almost their entire life without knowing that they have the disease. The mean, mode, and median of diagnosis all peak between 33 and 40 years of age, with another spike occurring during early puberty in the age cohort of 10-14 years of age (11.2\% of all diagnosed).


With the exception of the age cohort 25-29 years, the age of diagnosis between female and male shows a consistent pattern between the sexes throughout the age continuum. Within the age cohort of 25-29, there is a marked disparity, with five times the number of women (10.82%) diagnosed at that time compared to 2.2% of males diagnosed.

The possibility of a link between childbirth and the onset of CMT symptoms may clarify this disparity. The average age for first-time mothers in Australia has been steadily rising in this 25-29 age cohort; the average maternal age in 2010 was 30.0 years compared with 29.2 years in 2001, and 42.1% of first-time mothers gave birth at age 28.0. Focus group discussion covering the age of first diagnosis suggests that for some women, CMT was unnoticed before childbirth but became evident soon after. Word content analysis positioned the phrase ‘soon after (birth, first child, my first)’ at 19.5% when the topic of first diagnosis was addressed. Further, focus group participants diagnosed with CMT before the birth of their first child believe that their CMT symptoms worsened after the birth. However focus group data was non-conclusive on whether subsequent births worsened CMT symptoms.

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It was not methodologically possible to establish focus group data on the relationship between births after the first and worsening symptoms, but SCWM data confirms that 43.64% of women who have given birth believe that their CMT symptoms progressed soon after childbirth. A further 21.21% of mothers were ‘not sure’ if childbirth had made their CMT symptoms worse after childbirth.

Note: Type of CMT to age of diagnoses and SCWM frequency.
The most frequent type of the disease, CMT Type 1A, a demyelinating peripheral neuropathy characterized by slow nerve conduction velocity, distal muscle weakness, atrophy, and sensory deficiency.\(^\text{23}\) The average age of onset of clinical symptoms is 12.2 +/- 7.3 years.\(^\text{24}\) CMT Type 1A is a slowly progressing form of CMT with individuals typically exhibiting symptoms between the ages of 5 to 25. People with CMT Type 1A live a normal life span with fewer than 5% wheelchair dependent.\(^\text{25}\)

SCWM data shows the highest cohort (14.75%) of diagnosis for CMT Type 1A is age 45-49. The second highest cohort (7.38%) for diagnosis is 15-19 years of age. CMT Type 1A is not normally diagnosed until later in life for sporadic cases where no family history can be used as an indication; this is evident in the SCWM data with the 0-4 cohort at 4.10% and the 5-9 cohort at 2.46%. This is a contrast to the more severe form of CMT Type 3 Dejerine-Sottas, with a much earlier onset, which is reflected in the diagnosis figures from the SCWM. The majority (52.94%) of pwCMT Type 3 were diagnosed early in life (at 14 years or less). There were no cases of pwCMT Type 3 diagnosed after the age of 39 in SCWM data. The highest frequency cohort of diagnosis was 0-4 years of age (23.53%).

SCWM participants were asked if they had undergone definitive genetic diagnosis testing. Across Australia, the majority of SCWM participants (60.6%) had been genetically tested for CMT Type, 33.9% had not, and 5.5% did not know if they had been genetically tested. Focus group data substantiates that discovering for the first time that they have CMT can for many people be a confusing and stressful time.

“I know I have CMT but I am not sure which Type. There were so many blood tests and scans it was confusing.” Focus Group quote.

It is likely that this confusion had much to do with the 5.5% who were not sure if they had been genetically tested. Too many Australians with CMT do not know their CMT Type, with 36.1% either not knowing if they have been genetically tested, or who had been genetically tested and


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still were unaware of the type of CMT. To understand the CMT type is to assist with the correct treatment and management of CMT. SCWM data indicate that the decision for an individual to consent to genetic testing varies significantly from state to state and between major cities, rural and regional areas.

Availability and cost of genetic tests

Genetic testing for CMT type may differ according to where in Australia the pwCMT lives and the health care professional’s level of knowledge, commitment or funding. The cost of testing in Australia varies widely, conditional on whether the test is covered by Medicare, or if the individual is a private patient or a public clinic patient.

“In 2006, only 5 of the 437 tests available were covered by Medicare.”

Unconditional genetic testing for CMT is not at present covered by Medicare. Most tests offered through public clinics are wholly or partly subsidized, however long waiting periods for an appointment was a theme commonly expressed in focus groups. A subsequent concern was the unknown about genetic testing and insurance.

“The reason I have not been tested is because I would then have to declare a pre-existing condition. I will never get tested and my son will not either.” Focus Group quote.

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Such concerns expressed in focus groups may very well be justified, as a genetic test for CMT has implications for various insurance products. Insurance clients must declare factors affecting risk assessment, including family history or the result of genetic tests that the pwCMT or other family members have had. Failure to declare may invalidate the policy contract.

Privacy issues are reported to be of concern in regard to how genetic information is secured. In recent years there has been an increasing amount of proven genetic discrimination mainly to do with life insurance, and predominantly onset of illness and its impact on income polices. Insurance policies have been denied, or exclusions have been enforced in an arbitrarily far-reaching way given the narrow range of their genetic issues. The decision to genetically test or not is now becoming an economic one.

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Each pie chart plotted on the Australian map consists of three categories in answer to whether or not the SCWM respondent has been genetically tested for CMT type. Frequency of genetic diagnoses according to national and state location is examined further in sub-national jurisdictions, as well as comparative frequency over major cities and rural and regional areas.

The ACT has the highest percentage of genetically tested pwCMT (82%) of the states or territories in Australia. The ACT is also the only jurisdiction where SCWM participants knew for certain if they had been tested or not. It can be said however that while people in the ACT top the scale for understanding whether they have been genetically tested for CMT or not, it is still not ideal that almost one in five (18%) pwCMT in the ACT have not been genetically tested.
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PwCMT who reside in NSW have the second highest percentage of genetic testing (73%) in Australia, with 8% being ‘not sure’ whether they have been genetically tested or not. More people are genetically tested for CMT in Sydney than in any other area of Australia.

A closer look at NSW genetic diagnosis frequency location shows significant disparity between urban and rural Australia. The ABS Australian Statistical Geography Standard (ASGS) Remoteness Index plots all 148 NSW SCWM participants. Each NSW SCWM participant was classified into five different categories within the ASGS guidelines: pwCMT living in ‘major cities’ as opposed to those residing in ‘inner regional areas’, ‘outer regional areas’, ‘remote areas’ and ‘very remote areas’. In Sydney, 82% of pwCMT were genetically tested, with 12% not sure and 6% not tested.

Significant urban areas in NSW under the ASGS classification include Wollongong and Newcastle. Wollongong, a ‘major city’, also includes the Illawarra region along with neighboring Kiama and Shellharbour. Around Newcastle, the Hunter region includes Lake Macquarie and Maitland, classified as ‘major cities’. More pwCMT residing in major NSW cities are ‘not sure’ if they have been genetically tested than those who know if they have ‘not’ been genetically tested.
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The inland cities of Bathurst, Orange, and Cowra, as well as the northern city of Lismore, fall into the ASGS classification of ‘inner regional’, with Parkes ‘outer regional’ and Dubbo a ‘remote area’. More people have not been genetically tested in Lismore than those who have. The ASGS remoteness index indicates that CMT diagnosis frequency drops in line with the remoteness index.

![Genetically Tested for CMT in QLD](image)

Genetic testing in Queensland, in particular in regional areas, is difficult outside of the major city of Brisbane. Focus groups in regional Queensland (Dalby and the Sunshine Coast) reveal few neurological specialists willing to undertake genetic testing or referral to health care professionals. Several focus group participants reported traveling to Sydney for testing, or stated that testing was conducted previous to the pwCMT relocating to Queensland.

![Genetically Tested for CMT in SA](image)
With 53% tested, South Australia genetic testing sits at one of the lowest frequencies of any Australian state. Recent awareness campaigns promoted through a well-organized South Australian CMTAA branch is expected to increase frequency. The CMTAA national website has information to better inform pwCMT on their genetic diagnosis options.

Genetic testing in Tasmania at 37% is the lowest level in Australia. Focus group data revealed many pwCMT in Tasmania who have been diagnosed traveled to Melbourne for genetic testing.

“My GP thought I might have CMT and referred me to a neurologist in Hobart. It was 5 months later and $200 to see the neurologist for 10 minutes. There was no mention of genetic testing.”

The CMTAA in Tasmania provides members with up-to-date information regarding CMT diagnosis. Many focus group participants commented favorably on information provided by the CMTAA website. Regular CMTAA meetings in Hobart assist newly diagnosed pwCMT who often face long delays and poor communication with Tasmanian health care professionals.
At 57%, CMT genetic diagnosis testing in Victoria falls well below NSW. One regional Victorian focus group included a family of pwCMT who were unaware that the CMT disease (as they knew it) existed outside their family. This Victorian family attended to all their CMT secondary care almost completely autonomous of health care professionals. The family had shoes specially crafted, and made their own adjustments to motor vehicle controls as well as adjustments to heavy farm equipment. Another Melbourne focus group included a family of pwCMT of Vietnamese ancestry, unable to communicate in English, and who also had no understanding of the disease, even after consulting with a Melbourne neurologist.

“I received this letter. I do not understand what it means. Where do I go now?” Focus Group quote via Vietnamese interpreter.

In this case, the test results were mailed in English and no follow-up appointment was recommended. The necessity of improved communication between the newly diagnosed pwCMT and treating health care professionals is a common theme in focus group discussions.

PwCMT in Western Australia who have been diagnosed are well informed of their genetic type, with 72% being genetically tested and no SCWM participants who are not sure if they have been genetically tested or not. The CMTAA has regular support group meetings in Perth and health care professionals routinely attend.

A significant proportion of pwCMT in Australia do not know what type of CMT they have, with 43.2% of SCWM participants unable to state a type.
Reducing the Health Burden of Charcot-Marie-Tooth in Australia

Survey (SCWM) Percentage of CMT Type

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<tr>
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# Reducing the Health Burden of Charcot-Marie-Tooth in Australia

## CMT Type 3 Table vs. CMT Type X Table

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## CMT Type Other Table vs. CMT Type Not Sure Table

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Reducing the Health Burden of Charcot-Marie-Tooth in Australia

A closer examination of those people who did not know their CMT type shows 81% were first told that they had CMT by a neurologist, 6% by a General Practitioner, 7% by other parties, and 6% by a family member. Focus group data revealed that many had either forgot, expressed little interest or were not informed. Some people (5.09%) indicated that their type of CMT was still unknown.

Much of the self-reporting data presented in this section to do with diagnosis has been sourced from the SCWM and focus groups. Data collected from health care professionals confirms that while there has been progress with avenues for genetic testing there is substantial room for improvement. With CMT, 65% is due to known genes, the rest needs to be investigated by research methods anyway, hence slightly beyond the diagnostic testing conversation. Further, with CMT due to known genes, the advantage is that 60-80% is due to CMT1A (80% in children). This can be easily diagnosed on a Comparative Genomic Hybridisation (CGH) array, a test available throughout Australia. Hospital specialists can easily order this test, but private practice paediatrician and neurologists can only order this test for developmental delay, and not for diagnosis of CMT.

There are advantages to allowing private paediatrician and neurologists to request this test on a patient who has an obvious peripheral neuropathy that looks genetic, especially if there is a family history. This would typically shave eight months off the hospital process; the time it takes a hospital neuromuscular specialist to see the patient (usual waiting list six months) and get the result of the test (usually two months).

For the remaining 20-40%, Medicare does not fund genetic testing. The process of organising genetic tests here varies among paediatric and adult hospitals. Some hospitals have limited funds for such genetic testing with the requesting clinician needing to show immediate benefit (prenatal counselling, treatment modification) to qualify. However, the access to neuropathy panels in Australia allows for testing all known genes once funding has been arranged.

An important element to genetic diagnosis is its ability to lead to possible prevention of the disease. Some pwCMT in Australia have reported using a technique known as preimplantation genetic diagnosis (PGD). PGD is a technique that allows people who carry CMT to have treatment at IVF clinics to prevent passing on the condition. PGD uses standard in vitro fertilisation (IVF) during which eggs are fertilised by sperm in a laboratory. Embryos are
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matured and a sample of the embryo’s DNA is tested for the presence of genetic variation. Embryos that do not carry the genetic change are placed into the womb. The technique is able to detect the specific DNA change that initiates the genetic condition and is used to screen for an extensive variety of conditions. PGD is only possible if the genetic mutation is mapped, for which many CMT types have been.28

The decision whether to use PGD is an individual one. However the decision can only be made if pwCMT are aware that the science is possible to prevent passing the disease to the next generation. SCWM data tells us that 84.1% do not know what PDG is.

![Pie chart showing 15.9% have heard of PGD and 84.1% have not.]

Of the 15.9% of SCWM participants who have heard of PGD, just over half 53% would consider to use the technique, with another 27% not sure and one in five people would not use PGD. Focus group data identified several reasons why they would not use PGD, including the financial cost, ethical and religious beliefs, the nature of the procedure, as well as the perceived lack of likelihood of success.

“Is it true that it often fails? I know that it is expensive.” Focus Group quote.

In Australia, PGD must be funded for privately and one in three SCWM (34.2%) found that cost prohibitive. A significant 42.1% were not sure is the cost would be prohibitive and focus group data further confirms there was a great of confusion about the cost of PGD.

A PGD development, karyomapping, is now offered in Australian fertility clinics. Using DNA finger-printing technique to identify which embryos have inherited the chromosome carrying the altered gene, karyomapping may only take weeks to perform rather than older PGD techniques which can take several months. Techniques such as PGD and karyomapping provide options to pwCMT who may wish to avoid passing on CMT to their children.²⁹

3. Medication

The use of prescription medications can significantly improve the health and QOL for pwCMT. If used correctly, medications can assist in pain management as well as secondary conditions typical for CMT. Conversely, the misuse of medicines, either singly or in combination, may lead to reduced effectiveness, drug interactions, unpleasant side effects or more serious adverse reactions. In 2008, total expenditure on pharmaceuticals & other medical non-durables per capita in Australia was US$503.30

SCWM data has indicated that 71.6% of pwCMT used one or more prescription medication as opposed to 59.0% of the general population. Of those, 18.4% took five or more prescription medications on a regular basis. National Health Survey data found that the number of Australians using medications had declined from 64% to 59% after standardisation for differences in age and sex.31

SCWM data shows that 28% of pwCMT did not use prescription medication; 16% used one form of prescription medication; 17% two; 11% three; 8% four; 18% five or more. In comparison, 47% of the general population used one form of prescription medication, with 26% using two types; 12% three; 10% four; 5% five or more. There was a marked increase in the number of pwCMT using five or more medications (18%) compared to the general population (5%).

31 ibid
A higher proportion of females (64%) than males (54%) recently used a medication. This was more apparent in adults with the main differences between women and men evident in the use of pain relievers (27% compared to 20%). Amongst SCWM respondents, 83 females (43%) and 62 males (35%) were taking pain medication currently at the time of the survey.

SCWM and focus group participants most commonly reported using the pain medication Lyrica (pregabalin), an anticonvulsant that can affect chemicals in the brain that send pain signals across the nervous system. Another commonly used pain medication, Neurontin (gabapentin), also an anticonvulsant that has a similar mode of action as Lyrica. A significant number of focus group participants were using medications containing opioids to treat their pain. Both over the counter preparations like Panadeine (paracetamol and codeine), or prescription medications like Panodol Forte (paracetamol and codeine) or Endone (oxycodone), were commonly reported as used to treat pain.
According to ABS data, the proportion of people who used medications increased with age, from 42% of those aged less than 15 years, to 86% of those aged 65 years and over. The types of medications used also differed across age groups, in part reflecting the changing prevalence of illnesses with age.

SCWM data shows that 30% of pwCMT surveyed did not use supplements/non-prescription medications; 27% of pwCMT used one form of supplements/non-prescription medications; 17% two; 8% three; 7% four; 8% five or more. A further 70% of all SCWM participants used at least one supplement/non-prescription medication compared to 26% of the general population.\(^{32}\)

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Females were more likely than males to have used vitamins or mineral supplements, or herbal or natural remedies.

One in four SCWM participants (25.5%) reported experiencing negative side effects from prescription medication, compared to 4% of the general population. Focus groups participants frequently reported a ‘domino effect’ in the need to take one medication to alleviate the side effects of another medication. The most commonly reported side effect was fatigue, lack of concentration, digestive problems and loss of sex-drive. Many pwCMT indicated that they were on antidepressants and/or pain medication. Of these participants, many reported fatigue as a contributing factor in their need to change their type of employment and/or the number hours they worked.

Half of SCWM participants (49%) reported that medication side effects negatively affected their social life.
For many pwCMT the cost of medication can be prohibitive with 10% of SCWM participants reporting not having filled a script or had refused medication because of the financial cost.

Evidence suggests that factors exacerbating hardship for people with chronic illnesses included ineligibility for government support, co-morbidity, health service flexibility, and health literacy. Subsequently, pwCMT from culturally and linguistically diverse or Indigenous backgrounds, and/or not in paid employment, who take multiple medications experience greater economic hardship, and this in turn may further hinder the management of their chronic condition.

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Many medications used to treat pain and depression cause fatigue and interfere with a person’s capacity for work. Ironically, because of their work limitations, these people often struggle to find the money to pay for their medications. Additionally, many Australians ‘fall through the cracks’ when it comes to health care costs, being ineligible for subsidised medication and health care, but not having sufficient private insurance to pay for these medications and services. As the CMT population increases in age in line with the ageing general population, so will the burden of cost for medications and health care increase for the government.
4. Mobility

Compared to the general population, pwCMT display substantially lower quadriceps strength. A definite relation exists between quadriceps strength and timed motor performances to do with basic mobility requirements such as walking, squatting, kneeling and negotiating stairs.

The impact of balance and strength reduction on time taken tasks for pwCMT is considerable. Not only do pwCMT need to stop and rest more frequently, there is also a strong reduction of time taken.34

Evidence suggests that individuals with good mobility have fewer depressive symptoms than people who had mobility problems, irrespective of the level of physical activity.\textsuperscript{35} Consequently, it is imperative that pwCMT maintain the best level of mobility that their individual situation allows. Mobility aids play a large role to facilitate maximum mobility. SCWM data shows that one in three (33.7%) pwCMT use a mobility aid of some type.

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The more severe the core activity limitation, the greater the expected necessity for mobility aids and other devices. However, reported usage of wheelchairs, electric scooters, and walking frames was less than expected. SCWM frequency for wheelchair (9%) or motorized scooter (6.9%) was actually slightly less than suggested in the anecdotal literature. Dependence on these ‘high-end’ aids was again related both to age and core activity limitation. Walking stick and walking frame use was also highly age dependent, although leg/foot weakness was also implicated. This suggests that CMT related weakness might add slightly to the increase in dependence on common walking aids normally associated with ageing. Mobility aids have much to do with empowering the individual with independence necessary to perform day-to-day undertakings.\\n
A symbol of independence is closely tied to an individual’s ability to drive a car. Driving represents autonomy and can allow easy access to social connections, health care, shopping, activities and paid employment. For a person with reduced mobility, driving increases the QoL in many areas.

Driving beyond one’s ability brings an increased safety risk or even life-threatening situations to all members of society. No specific studies have been conducted on the topic of CMT and driving, however there is convincing evidence (insurance company and state government data) that disabled drivers are as safe, if not safer than the general population.\\n
SCWM data shows that of the 16.2% who do not drive a motor vehicle, 70.5% do not drive due to having CMT. The choice to drive or not was one made in the main by the pwCMT.

“It just didn’t feel safe for a whole bunch of reasons. At the end of the day it was my decision....”
Focus Group quote.

While just over half (52%) of SCWM participants reported never regularly relying on others for transports, of this group 96% reported to still driving. For pwCMT driving is more than just a symbol of independence, it represents genuine independence. Driving a car is for some a prerequisite to maintain paid employment; as either a requirement of the job, or as the only practical means to commute to and from work.
Australian state governments offer a taxi subsidy scheme for people with disabilities. In NSW, the Taxi Transport Subsidy Scheme (TTSS) provides subsidised travel, allowing approved participants to travel by taxi at half fare, up to a maximum subsidy of $30 per trip.\(^{38}\)

Regrettably many pwCMT do not qualify for the TTSS (including other similar state schemes) eligibility requirements with some forced to quit paid employment due to TTSS ineligibility.

“I am a registered nurse. I had to leave the hospital because it was no longer safe for me to drive. I did not qualify for the taxi scheme. It was $80 a day to get a taxi and I just could not afford it. Tried to car pool but that did not last long.” Focus Group quote.

There appears to be a gap in the eligibility requirements which many pwCMT fill, along with subsequent labour force participation loss.

5. Sleep and fatigue

Sleep disorders and fatigue are an under-recognised consequence of many neurological conditions including CMT. These sleep disturbances, and accompanying fatigue, are both primary and secondary in their aetiology. SCWM data shows a high incidence of sleep disturbance in pwCMT.

A 2009 German study looked solely at the relationship between fatigue and CMT. This study found the prevalence of impaired sleep and excessive daytime sleepiness extraordinarily high in self-reported CMT patients, with no significant differences between the various subtypes of CMT. The study investigated the prevalence of fatigue, daytime sleepiness, reduced sleep quality, and restless legs syndrome (RLS) in 227 adults with CMT and found a significant impact on QoL. SCWM and focus group data corroborates these findings.

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Reducing the Health Burden of Charcot-Marie-Tooth in Australia

SCWM participants reported high levels of sleep disturbance, including heavy snoring (21.9%), feeling tired and unrested in the morning (48.5%), waking early (31.2%), waking in the night (61.1%) and difficulty getting to sleep (40.3%).

According to the German study, pwCMT reported significantly higher fatigue, a higher extent of daytime sleepiness and worse sleep quality. Prevalence of RLS was 18.1% for pwCMT and severity was correlated with worse sleep quality and reduced QoL. Women with CMT were affected more often and more severely by RLS.
One third (33.3%) of SCWM participants reported that they were either somewhat or very likely to fall asleep during the day. SCWM Australian data echoes the German findings which found that daytime sleepiness was reported by 31.7% of pwCMT, and that 79.3% were found to be ‘bad sleepers’. A Pittsburgh Sleep Quality Index (PSQI) global score >5 was found with a prevalence of 80.4% in the CMT1 group, 72.2% in the CMT2 group, 88.9% in the CMTX group, and 80.0% in the ‘other/unclassified CMT’ subgroup.40

The frequency of RLS amongst pwCMT in the study was 18.1% compared to 5.6% of the control group. The prevalence of RLS was 22.3% in the CMT1 group, 19.4% in the CMT2 group, and 12.9% in the ‘other/unclassified CMT’ subgroup. Only one out of nine CMTX patients had RLS. The mean onset of RLS symptoms was 27.7 years of age, with a range from 4 to 61 years. RLS was most common in participants above 60 years of age (23.5%), compared to 21.3% in the 30–60 years group and 16.7% in patients under 30 years. Women are more likely to suffer RLS than men with women making up the majority of pwCMT suffering RLS (68.1%). The study reported severe fatigue in 64% of 137 CMT1A patients and 43.2% of all survey participants.

Twice as many SCWM participants reported regularly suffering from physical (71.8%) than mental (34.2%) fatigue. While age can be correlated with fatigue in the general population, age is less of an indicator of fatigue for pwCMT.

Little is known about the mechanisms by which peripheral neuropathy leads to fatigue. Some people report fatigue in the presence of rather mild neurological disability. As it is for pwCMT, similar reported frequency between the different types and core activity limitation severity.

The relationship between CMT and fatigue is not attributed to poor sleep or daytime sleepiness. It has been postulated that fatigue and daytime sleepiness are different entities both caused by non-restorative sleep. Difficulties in falling asleep and frequent awakenings appear to be the most important reasons for non-restorative sleep in CMT patients.

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While it is difficult to specify exactly how many Australians suffer from sleep apnoea, evidence suggests that it may affect 4.7% of the general population. The prevalence of obstructive sleep apnoea (OSA) is increasing in Australia, probably due to the 'obesity epidemic'. Despite this, a large proportion of Australians with OSA remain undiagnosed and untreated.43

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PwCMT should be examined for sleep-related symptoms and fatigue in order to identify treatable conditions. Apnoea screening is appropriate in people with at least moderate disability, and Polysomnography should initially be performed if severe RLS, periodic limb movements of sleep, or if nocturnal hypoventilation is suspected. Since causative treatment for CMT is not available, sleep-related symptoms should be recognized and treated in order to improve QoL.  

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6. The relationship between pain and CMT

Health care professionals are divided on the relationship between CMT and pain. The study examines the frequencies, location, severity and types of pain, and how pain interacts in the social and working lives of pwCMT. It also examines the economic cost of pain. The extent to which pwCMT suffer pain has been the subject of a long debate among the medical fraternity. Our data collates the SCWM of 418 participants and 21 focus groups comprising 178 individuals to suggest that for those who suffer pain, it is a tough road.

Type of pain

There is an abundance of scholarly work covering numerous aspects of CMT, but there is very little on the relationship between pain and CMT. Anecdotal evidence from health care professionals and CMT support groups (CMTAA, CMTA) suggests that many pwCMT suffer pain. Two international studies, one from Italy (2008) and another from the United States (1998), demonstrated a link between pain and CMT. In Australia, SCWM data also supports this relationship between CMT and pain. CMT related pain falls into two categories:

- Neuropathic
- Postural related, as a consequence of musculoskeletal abnormalities developing (often over time) as a corollary of CMT.

The most common form of pain experienced is neuropathic pain, identified in the SCWM as including any or all of the following types: shooting, numbness, tingling, or burning. Neuropathic pain is present in 91.34% of pwCMT who suffer pain (neuropathic or postural related) and varies in frequency. While neuropathic pain is present as a symptom of CMT, it is comparable to that seen in other peripheral neuropathies, as well as post herpetic neuralgia and complex regional pain syndrome, among other diseases. Non-neuropathic CMT pain is invariably due to postural changes, musculoskeletal abnormalities, scoliosis, foot deformities, fatigue and cramping and this type of pain is common. Of the 278 SCWM participants who indicated that they had pain, 67 had cramping or aching, while 24.10% with pain had non-

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47 Ibid.
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neuropathic pain. Of the 24.10% who indicated postural related pain, 67.16% also suffered from neuropathic pain.

SCWM and focus group data identifies the frequency and severity to which people reported pain and how this pain interfered in their social and working lives.

“Neuropathic pain is a significant problem for many people with CMT. Further studies are needed to examine possible pain generators and pharmacologic and rehabilitative modalities to treat pain in CMT.”\(^{48}\)

**Pain frequency and severity**

Of the SCWM participants, **71.06%** reported pain. In the early onset group with observable symptoms 0-9 years of age) who reported pain, a higher frequency of pain was reported (77.74%) as opposed to 64.49% of late onset (observable with symptoms = >10 years of age) pwCMT. Women reported slightly more pain than men in each of the three categories (general CMT, early onset CMT, late onset CMT).

Of the pwCMT surveyed who indicated that they experienced pain, whether neuropathic or postural, in the last four weeks, 39.2% rated that pain as moderate. A further 22.2% rated their pain as severe or very severe. Some studies on pain have used composite neuropathic pain measurement tools, such as the Neuropathic Pain Scale (NPS). The NPS was not adopted for the SCWM as it was judged problematic when presented in the form of a self-reporting survey. The SCWM did however use a version of the NPS scale, where participants were provided with a 5-point scale of severity (None, Very Mild, Mild, Moderate, Severe, and Very Severe) rather than the NPS 10-point scale (Intense, Sharp, Hot, Dull, Cold, Sensitive, Itchy, Unpleasant, Deep, and Surface). To ascertain how pain relates to social and working lives, SCWM participants were asked to identify the type of pain they experienced from five categories (Burning, Tingling, Shooting, Numbness and Aching) and to plot each category from eight body locations (Lower Legs and Feet, Hips/Thighs/Knees, Lower back/abdomen, Torso/Upper Back, Forearms/Hands, Shoulders/Top of Arms, Neck, and Head/Face).


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“All the time I have burning feet. They keep me awake all night and I can’t stand it anymore. My doctor shrugs it off as if I’m making it up.” Focus group quotation.

Burning pain was most frequent in the lower legs and feet as well as the forearms and hands. Of the 71.06% who suffer pain, 42.45% experienced burning pain in the lower legs and feet and 19.42% in the forearms and hands. Qualitative focus group data indicated that pain might also have diverse time qualities. Some people experience stable pain that really does not change in severity, whereas for others the pain is intermittent, with intervals that are entirely pain free. Others are never without pain, but their pain severity is variable from one instant to the next. For one in three (33.80%) of pwCMT who suffer burning pain in the lower legs and feet, this pain is severe to very severe. At times, intensification can be severe with periods of very severe pain. At other times individuals can feel lesser levels of background pain. Nevertheless, they report never being free of pain.
“It’s strange, I can’t feel my feet most of the time. But if I walk without shoes on a rough surface I will be up all night with shooting pains.” Focus group quotation

Of those who reported pain, 57.91% experienced numbness in lower legs and feet. Focus group data revealed that for many, numbness was mixed with severe shooting pain. Hyperesthesia, numbness and shooting pain in the feet were reported in 18 of the 21 (86%) focus group discussions. More than three quarters (76.87%) who suffered numbness also suffered shooting pain in the feet. Almost half (48.39%) experienced severe to very severe numbness in the lower legs and feet. Almost one third (31.39%) reported pain had numb forearms or hands. Focus group word content analysis reveals that of this 31.39% group, most had numb fingers (95%). Further, the main issues for pwCMT with numb fingers were writing (85%), doing and undoing buttons (85%), feeling for objects in the dark 80% and burns/scalding (45%).
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“Tingling for me is like a constant reminder something is wrong. My feet usually start to tingle just before the painful shooting starts.” Focus group quotation.

Of the 71.06% of pwCMT in SCWM data who suffer pain, 34.17% reported tingling in the lower legs/feet and one third (33.33%) judged this tingling to be severe to very severe. A further 26.99% reported tingling in the forearms and hands, with 19.33% describing tingling as severe to very severe. The frequency of hips/thighs/knees tingling stood at 12.33% with a drop off on severity as 42.86% reported a mild sensation. While 60% reported severe to very severe tingling in the neck, only 1.98% reported neck tingling. The high severity of neck tingling can therefore be ruled out.
As a peripheral neuropathy CMT affects both motor and sensory nerves more the further the distance from the spinal cord. The location on the body of shooting pain frequency appears to follow the same peripheral nature of the CMT neuropathy. Thus shooting pain for pwCMT is most frequent in the lower legs and feet (34.17%). In regard to the hips/thighs/knees, the frequency reduces to 19.06%, then falls off to 8.99% in the lower back & abdomen and 4.31% in the torso & upper back. Hyperesthesia shooting pain was extensively reported in focus group quotations.

“The problem with Lyrica and other nerve pain medication I’ve tried, is that I need to take it every day, yet I don’t have the pain every day. The side effects knocked me about so much I had to quit my job.” Focus group quotation.

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discussion as the sensation that interfered the most in the day-to-day lives of pwCMT. The severity of shooting pain was greatest in the lower legs and feet, with 36.37% noting severe or very severe pain. In the same way that pain frequency reduces the further up the body towards the spinal cord, so too does the severity. Severe or very severe hips/thighs/knees was reported by 19.35%, lower back and abdomen by 8.33%, and 11.11% reported severe pain in the torso & upper back, with no reported cases of very severe pain.

"My hands and wrists ache most of the time and doing even basic tasks can be difficult. Along with reduced strength in my hands, I am very limited by the pain." Focus group quotation.

Focus groups assisted in clarifying what brought about the aching pain reported by so many in the SCWM. Postural/skeletal changes to the body as a result of CMT contribute to the majority of aching pain. For the 33.81% of pwCMT who reported aching lower back, many had a degree
of scoliosis. A high percentage (49.24%) reported suffering from arthritis. However, the most frequent aching pain (58.63%) was reported to be in the feet, associated with pes cavus (high arches) and/or hammer toes (a typical symptom of CMT). High walking gait and abnormal walking posture is linked to hip dysplasia, with 42.81% reporting hip pain.

**Pain treatment**

Focus groups offered a unique source of data collection using a flexible qualitative approach to pursue the pain topic in ways where the SCWM could not. Focus group participants were asked open-ended questions to expand on areas already identified in SCWM data; the extent to which pain interfered in their day-to-day lives and how health care professionals treated their pain.

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How well have your health care professionals treated your pain since it first manifested?
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<table>
<thead>
<tr>
<th>Response</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Not at all</td>
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</tr>
<tr>
<td>Poor</td>
<td>21%</td>
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<tr>
<td>Fair</td>
<td>24%</td>
</tr>
<tr>
<td>Well</td>
<td>17%</td>
</tr>
<tr>
<td>Very well</td>
<td>34%</td>
</tr>
</tbody>
</table>

“Persistent pain management services are currently unable to meet service requirements adequately, and waiting times are more prolonged for publicly funded than privately funded services. Greater service provision is required in rural areas and for children.”

For those participants in the focus group who were not satisfied with the way in which health care professionals treated their pain, issues surrounding health care professional scepticism were common. Many reported that even when receiving a script for a narcotic or opiate drug, there was a constant questioning of whether or not they were doing something wrong. This

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would not only happen at the GP level but also at the time of filling the script at a pharmacy. A number of pwCMT reported pharmacies refused to fill scripts.

“In April I saw in the media that over-the-counter codeine and other some pain relief medication may be stopped. I actually thought of going down and stocking up. Not getting pain relief terrifies me.” Focus group comment.

Most focus group participants reported that specialists understood their pain better than other health care professionals. The limitation of existing treatments for neuropathic pain and the incapability to provide respite for many patients has driven ongoing studies on diverse approaches to averting neuropathic pain.53

Across all types of CMT, 17.5% of SCWM participants reported scoliosis. Comparative international studies on the CMT-scoliosis relationship report a higher frequency (33.33%), with the average age of diagnosis of spinal deformity as 12.9 years. Surgery intervention was performed in 31.11% of patients.54 Surgical intervention associated with scoliosis was 15 out of 70 (21.43%) across all CMT types.
Of the SCWM participants who indicated that they suffered scoliosis, the severity of pain drifted towards the higher end of the severity scale. When comparing pain severity data between individuals who suffered mild or very mild aching pain (56.25%) with those who have scoliosis, many reported mild or very mild aching pain dropped to 28.23%. When comparing pain severity data between all pwCMT who suffer severe or very severe aching pain (17.07%) against those with scoliosis, the scoliosis cohort with severe or very severe aching pain increases to 46.56%. Similar upward trends are evident in comparative shooting pain data between all pwCMT and those with scoliosis. PwCMT and scoliosis are more likely to suffer shooting or aching pain in the lower back. Focus group data points to this issue as the major factor in people withdrawing from paid employment, whether office or manual work.

The percentile range of reporting pain between the different types of CMT was 65.38% for CMT2 to 84.47% for CMT type 3. CMT type 3, Dejerine-Sottas Syndrome (DSS), is a particularly severe variant of CMT with profound core activity limitations developed since infancy.

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Pain and depression

"Depression costs the Australian economy approximately $12.6 billion per year and accounts for up to six million working days of lost productivity".\textsuperscript{56}

The frequency of pwCMT in Australia suffering depression is staggering. Many reported suffering from depression (47.66%), with 15.89% not sure, compared to 6%\textsuperscript{57} of people within the Australian general community with depression.

When looking at pwCMT who suffer pain, the frequency of those who report depression is even greater at 54.74%, with 16.06% not sure. Evidence suggests that 40% of those with depression seek diagnosis and subsequent treatment.\textsuperscript{58}


\textsuperscript{57} Gender Indicators, Australia, Jan 2013. 4125.0[ONLINE] Available at: http://www.abs.gov.au/ausstats/abs@.nsf/Lookup/4125.0main+features3150Jan%202013. [Accessed 29 September 2015].

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SCWM data supports this picture, with 20.70% of pwCMT formally diagnosed with depression. Again, this figure increases for those who have pain, with one in four (26.26%) with pain also having depression. The ABS National Survey of Health and Wellbeing notes that 6.2% of adult Australians have ‘affected disorders’ and 4.1% have ‘depressive episodes’. Of Australians aged 16-30 years, 90% are employed and/or enrolled in study towards a formal secondary or tertiary qualification, but this drops to 79% for those who suffer a mental illness.

Anxiety disorders and pain

In Australia, anxiety disorders are the most common mental disorders, affecting 14% of all people aged 16-85 years in a twelve-month period. Anxiety disorders commonly encompass thoughts of tension, nervousness or distress. Certain anxiety disorders such as panic disorder, agoraphobia and generalised anxiety disorder have similar symptoms such as problems in breathing, heart palpitations, perspiring, trembling and shaking. Within the general population, 18% of women experience anxiety disorders compared to 11% of men, and women aged 16-54 (21%) experienced anxiety more frequently than older women aged 65-85 (6.3%).

Anxiety self-assessed: pwCMT, pwCMT and pain, general population

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For pwCMT, anxiety is much more frequent than in the rest of the Australian community, with 38.6% of participants reporting suffering from anxiety, and even more frequent (44.8%) for those who suffered pain.

Focus group data identified two common themes when examining the relationship between pain, CMT and anxiety. First, pain management had much to do with anxiety. PwCMT exhibit anxiousness over their perceived health care professionals’ lack of understanding with pain relief requirements, such that acquiring medication over a long period of time to treat their pain was problematic. This was further compounded for those who did not have a regular GP who understood their medical history best.

“My doctor sometimes gets the idea that he should lower my dose, or change the medication altogether. I am terrified that he will one day say no to what I know works. And if I am forced to see another doctor from scratch to prescribe my medication, it will be very difficult.”

The second common theme presented in focus groups to do with pain and anxiety was pain itself. Pain may appear at any time and this contributed to anxiety. SCWM participants indicated a degree of anxiety (23.28%) about leaving the home, with 3.70% not sure. Word content focus group analysis indicated that 15% of the 23.28% who did not leave the home did this because of experiencing pain, or the fear that pain might occur when out. Focus group data further revealed that many felt anxious about leaving the home because of CMT. PwCMT found it more difficult to walk or mobilise outside the boundaries of their home. Trips and falls are common, and uneven or loose surfaces increase the chance of falls and perhaps injury. Tripping (96.1%), the need to be cautious when walking (96.1%), and difficulty with slippery surfaces (95.8%) are everyday hazards.64

“I have to plan all my trips. Even going to the shops is an effort. I get anxious if I can’t get the car park I like. I miss out on countless outings because I get anxious about my mobility.” Focus Group quote.

Just over half of pwCMT who believe that they suffer from anxiety have been diagnosed. Of all pwCMT, 21.7% have been diagnosed by a health care professional and for those who also suffer pain, 25.2% have been diagnosed as suffering anxiety.

Living with pain

As there are no known disease-modifying treatments for CMT, those who suffer CMT-related pain typically endure it for the rest of their lives. CMT-related pain is unique to each individual. The nature of both neurological and CMT postural related pain may be intermittent, often interrupted by gaps where pain is absent altogether, or varies in severity. Focus group data suggests while pain severity also fluctuates, it does not withdraw altogether. Across all pain
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categories, participants reported it was often worse at night just prior to sleep. Burning pain however was more common in the morning, soon after or immediately after waking up. Accordingly, CMT pain falls into the category of chronic pain. The vast majority (85%) of pwCMT who suffered chronic neuropathic pain expressed frustration at the random nature of where, and more so when, pain would occur. This randomness was felt most in the working lives of pwCMT. Chronic pain has a significant economic impact on the economy in both prevalence and the wide-ranging and substantial influences on pwCMT themselves.

Cost of chronic pain

“The total cost of chronic pain in 2007 was estimated at $34.3 billion – or $10,847 per person with chronic pain.”

The pro-rated estimate based on the cost of chronic pain within Australia as of Dec 2007 is $64,238,103. The national cost of pwCMT who suffer chronic pain, broken down by each state and territory, does not include the burden of disease (BoD) estimates and should be viewed as purely pain-related costs.

<table>
<thead>
<tr>
<th>Location</th>
<th>Total</th>
<th>PwCMT</th>
<th>PwCMT &amp; pain</th>
</tr>
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<tbody>
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<td>$21,132,125</td>
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</tr>
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</table>

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The distribution of CMT pain costs is shared between the federal and state governments, the individual with pain, society, family and friends. Government bears the majority of this cost, in a ratio of 45% by the federal government to 22% by state government.

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The main portion is productivity costs (34%), a strong indication of the significant impact on work performance and employment outcomes in those affected by chronic pain. These work performance outcomes are evident in the SCWM data related to participant perception of how pain frequency and severity had much to do with the ability to complete work tasks satisfactorily and on time.

It is the individuals themselves who must endure the main burden of chronic pain costs, mostly because of the BoD costs (55%). The Federal Government, in particular the health system and productivity, costs 22%, followed by state government (5%), employers (5%) and then society in general, family and friends (13%).

“Interventions that target working with appropriate pain management, together with other support such as job flexibility, could significantly reduce lost productivity costs due to chronic pain.”

Just under three-quarters (74%) of pwCMT who suffer pain believe that pain interfered with their capacity to work at some level. Chronic pain has a substantial bearing on a person’s ability to work efficiently. For some (16%), chronic pain was the reason to cease work. Many factors have the potential to inhibit the recruitment and retention, further enhanced for pwCMT with chronic pain. Focus group data has identified the following issues;

67 ibid.
68 ibid.
Pain is invisible and there is little information and support about working with pain.

As there is little existing evidence about the relationship of CMT and pain, many reported that there is a stigma attached to declaring their pain-related work issues.

Many hid their pain to avoid workplace discrimination and in turn compounded the problem.

An added impediment for pwCMT who suffer chronic pain is the extra effort, time, and cost in commuting to work as some pain medications restricted driving.

Side effects of pain medication either interfered or restricted totally the individual’s function in the workplace. Some reported having to avoid taking the ‘best’ medication to treat their pain (neuropathic or CMT postural related) because of their unique workplace requirements.

PwCMT also reported that employers expressed apprehension about workers compensation liability. Consequently, a number of those who reported chronic pain have splintered working lives, unable to maximise their earning capabilities by way of promotion restriction or ability to take overtime.

Pain self medication
For most pwCMT (71.2%), their use of alcohol is moderately benign. A small proportion of those who do drink, do so to relieve diagnosed or undiagnosed depression, anxiety or physical pain. Of the 28.8% who do consume alcohol, 20.1% drink once a month or less. A further 18% drink a few times a month and 16.7% drink a few times a week. However, 17.7% drink every day and 10.2% five drinks or more a day. While the vast majority drink moderately, 8.4% consume six in one sitting a few times a week or more. For some pwCMT drinking is a way to self medicate.

“It is easier for me to get pain relief from the bottle shop rather than the doctor who I felt just didn’t believe me. The bottle shop employee was not as judgmental.” Focus Group quote.
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“*I often reach for a drink to calm my nerves or to relieve my emotional pain.*”

Focus group quotation.

There is evidence that alcohol escalates an already high frequency of depression and anxiety in the CMT community. Alcohol may also affect the efficacy of antidepressant medication. Focus group data found that rather than seek out help to manage depression or anxiety, some pwCMT sought their own solution, a solution which may work with a perceived success for a while, but ultimately complicates the issues and leads to more emotional pain. A number of pwCMT consumed alcohol to assist with their physical pain.

However there is strong evidence that the abuse of alcohol is more damaging for CMT nerves which already display a degeneration secondary to the undetermined genetic defect. 11.2% of SCWM participants reported injuring themselves while under the influence of alcohol. Focus group word content analysis indicates that 42% of pwCMT alcohol-related injuries required medical attention. Alcohol-related harm results in 65,000 hospitalisations every year in Australia. The direct cost of alcohol related injury and problems to Australian society in 2010 was estimated at $14.352 billion.

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The burden of alcohol costs is divided between three segments of society: families, business and government. Business has the greatest proportion of the burden with 50% of the costs, followed by the government sector with 26% and families with 24%.\(^{73}\) Much of the cost to industry is due to reduction in the volume and capacity of the workforce and worker absenteeism because of alcohol-related issues.\(^{74}\)
7. Caring for people with CMT

In line with government policy of the past three decades, and the ensuing movement away from institutionalised care in the direction of community care, family members, partners, close friends and even neighbours, now perform the responsibility of care. For many people, caring can be both rewarding and fulfilling. However it can also have an impact on the carer’s own health, both physical and mental, and may also have an impact on their capacity to pursue their own life goals of relationships, travel, employment and education.

In 2012, there were 2.7 million carers in Australia, representing 11.9% of the population. For the narrower purposes of this study, a carer is an individual of any age who cares for a pwCMT who has limitations in performing core activities, self-care and mobility. This care is ongoing and unlikely to cease due to health improvement.\(^7^5\)

SCWM data confirms that one in five pwCMT (21.5%) require a carer, and that more than nine out of ten carers (92.68%) live in the same household as the person for whom they provide care. For pwCMT, the severity of disability can be measured by the degree to which they are restricted in performing core activities. For the purposes of this report, the Australian Bureau of Statistics Core Activity Limitation scale has been used to distinguish four levels of disability: profound, severe, moderate and mild (see section 1.5.1).

Caring for a child, partner or parent with some degree of core activity limitation is mainly a family role that many people may perform during the course of their lifetime, with the focus of care changing as people grow older. Overall, women are more likely than men to be a carer (14% compared with 11%).\(^7^6\) Across the entire Australian carer community, 43% of primary carers were caring mainly for their partner, 24% for one of their parents, and 22% for one of their children. Carers aged 15-34 years were most likely to be parents caring for children (35%), while those aged 35-64 years were most likely to be caring for their partner (36%). Three quarters of primary carers aged 65 years and over were caring for their partner.\(^7^7\)

The probability of a carer living in the same household varies according to the relationship of

\(^7^6\)Ibid.
\(^7^7\)Ibid.
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the carer to the main care recipient. Most of those caring for their partner or child were living with that family member.78

SCWM data indicates that the role of the carer follows two distinct paths. The first path, for people with late onset CMT, falls in line with societal expectations of the carer’s ‘focus of care changing as people grow older’.79 This path is typical for pwCMT with a mild or medium core activity restriction, often not diagnosed until the second decade of life and onwards. The second path, early onset CMT, follows a harder route, with profound or severe core activity limitation that often manifests itself from birth to ten years of age. For these people the level of care required increases both in daily tasks and in duration of care. As pwCMT typically live a normal lifespan, this care may extend to several decades or more.

An added difficulty for intergenerational care is the hereditary nature of CMT. One in every ten (10.49%) pwCMT are being cared for by another family member with CMT, with 29.27% caring for an adult, and 70.73% of care for a child. Of those who care for a child, 57.14% care for two or more children.

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SCWM data indicate that 76.81% of pwCMT have children with the early onset group less likely to have children (57.69%) than late onset (81.42%). PwCMT are more likely to stay in the family home longer than people without CMT. The 19-28, 29-38 and 39-48 age cohorts show a trend towards staying in the family home. However older people with CMT (49 plus cohort) are returning to the family home if the challenges of independent living become more apparent.

“I moved out of home in 1988 but I am back at home now because my CMT got a lot worse.”
Focus Group quotation.

Across all age cohorts, children of pwCMT who themselves have CMT stay in the family home longer than the general community.

In regard to people without CMT living in a household where one parent has CMT, focus group data shows two divergent trends. Some children without CMT felt obliged to stay in the family home to care for their parent, whereas other children felt as if moving out of the family home assisted their parent by reducing the burden of care they placed on the parent with CMT.

“Mum and I both have CMT. I am able to do more about the place than her these days. We often joke about it, how not so long ago she was looking after me, now it’s me looking after her. I had to quit my job last year though, so now I’m on the carers pension.” Focus Group quote.
Focus group data was polarized, with some carers expressing their role as a positive experience whereas others saw it as an arduous task. Nevertheless, all carers interviewed indicated a sense of fulfilment, but 60% stated that the financial and health costs weighed heavily. Australian carers are 65% more likely to reside in a family with a gross household income in the lowest two quintiles, compared with 36% of non-carers. Only 7% of carers had household income in the highest quintiles of gross household income, compared with 23% of non-carers. Being a carer may result in physical and emotional impacts with 50.4% having often felt weary, angry, resentful, worried or depressed. Notwithstanding these negative impacts, 27.8% of carers reported that they felt satisfied.

More than three quarters of pwCMT are in a relationship. For many their partner is also their carer.
“I am not sure if I am his lover or his carer [laughs]. We sure did start out as lovers and we still are. It’s just that these days I have to do so much for him. The CMT stops us from doing so much of what we were once able to do.” Focus Group quote.

When the carer of a pwCMT is the spouse or partner, the roles between carer and spouse are blurred. Of all those in care, 75.9% are cared for by their spouse. When excluding single people, the percentage of the spouse as carer increases even further. Thus a pwCMT who is in a relationship and being cared for is likely to be cared for by their spouse (91.43%), with the remaining 8.57% being cared for by children, parents, and unrelated persons.
Nearly half of SCWM participants (48%) believed that having CMT put pressure on their current or former relationships, and 20% believe that CMT was the primary reason for the breakdown of a previous relationship.

Carers who were in a relationship with pwCMT mostly believed that this dual role put pressure on their relationship, although some felt that the role bought them closer together. One in three (31.5%) of carers for all people needing care in Australia see the caring role as something that brought them closer to their partner. Half of the carers who are partners reported that their relationship was strained (50%), with 25% believing the role had been brought them closer together and 25% saw no effect.

“I had no choice to quit. My company was not able to give me the flexible hours I needed. Anyway we get to spend more time together although we miss the money. People say the pension pays well, as if. I was paying more tax every fortnight than what I am now earning. Sure, I would keep working but I can’t.” Focus Group quote.

Carers of pwCMT are more likely to be reliant on government pensions and allowances than the general population. The Centrelink Carers Payment Pension was the primary sources of income for 82% of carers aged 18-64 years, with 91% claiming the smaller Centrelink Carers Allowance. For those claiming a Carers Payment, the time expended in caring equated to that
spent on a full-time job. Focus group data shows some carers who have left paid employment to take on the caring role are highly skilled and educated and would otherwise be still engaged in paid employment.

For those individuals with a profound core activity restriction, the caring role required more. Carers of pwCMT with profound or severe core activity experienced reduced prospects of employment, especially full-time employment. Both focus group and SCWM data indicate that the time spent caring had much to do with a carer’s inability to participate in paid employment. Only one in every eight carers (12.5%) of those 15-64 years of age claiming the Carers Payment participated in paid employment. Carers overwhelmingly rated fatigue (45%) as the greatest hindrance to seeking paid employment, followed by lack of time (28%), and inability to find suitable employment that accommodates their role as a carer (19%).

For many carers (36.59%) the difficulties in maintaining paid employment and taking on the important role as carer necessitated changing their employment situation or conditions. SCWM data shows that 9.47% of pwCMT have had to change their own employment to help other person in their family who has the disease. Focus group data shows that of these carers who changed their employment conditions, 36% withdrew from the labour force altogether and a further 25% reduced their hours.

Nationally, more than half (58.2%) of carers do not participate in the labour force and more than half (54.9%) of primary carers reported a government pension or allowance as their main
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source of income. Many carer partners of pwCMT reported that their caring responsibilities affected them financially, with 40% reporting decreased income.

Over one in two (55%) carers for pwCMT required regular time off from work to manage their caring role, compared to 33% of all Australian carers. This is due to reduced options for outside assistance (2%). Most carers in focus group discussions expressed the hope that the NDIS would assist in providing outside help to avoid missing paid employment options.

The following case study looks at living with CMT from the carers’ perspective. The subject is a middle-aged woman who does not have CMT, however her husband and child have the disease. It is hoped that her insight will contribute to better understanding of the needs of carers.

I am a wife and mother and both my husband and my son have CMT.

I first learnt of CMT when I met my husband. I noticed his strange gait and that he had little muscle on his calves. He was very conscious of his feet so he never let me see them until well into our courtship. I never asked why and until we married, he never told me. I learnt of his condition through our doctor as my husband felt I needed to be informed due to the chance of our children inheriting the condition. As my husband’s two other children to a previous relationship appeared not affected, I did nothing more to learn about the condition. At that time he only occasionally needed assistance to open jars or help him up after a fall.

Over time it became more apparent that his symptoms were worsening so we relocated to a regional area to be closer to my support network from the family. My husband had stopped work and was becoming more frustrated with his growing weaknesses, so he often appeared rude and in a bad mood. He was also starting

84 ibid.
to withdraw more from socializing. He was becoming possessive of my time, making it difficult for me to have contact outside of the home.

This behaviour directly affected my ability to find help to care for him whilst I remained at work. Being closer to my family meant I could continue to work as there was support nearby if it was needed. This failed, as my family didn’t know about CMT and just thought he was lazy and unsociable which over the years has led to many arguments and has caused fragmentation in my relationship with both the family and my husband. I now attend all functions including family alone with my son to try and keep some sort of peace.

My son missed most development milestones and was diagnosed with Global Development Delay (GDD). He was not able to walk unaided and fell easily. Both of my GP and paediatrician were not up to date on CMT and fobbed me off as neurotic mum and told me that CMT could not be diagnosed until adolescence. I began looking up CMT to learn more about the condition as I knew in my heart my son’s issues were not just related to ‘all kids develop at different rates’. When he went to his first physiotherapy appointment it was noted how poor his balance was and immediately asked to see his feet. I mentioned his father has CMT and the physiotherapist arranged a referral for a neurological assessment. This led to his diagnosis of also having CMT1A just before his fourth birthday.

I had to give up full time work and become full time carer to both my husband and son to ensure my son could attend all necessary appointments. My husband’s pain, depression and fatigue were all increasing.

It was at this point I started to really try and find information on CMT so I could be more aware of what to ask for in his plan when the NDIS trials began. My son was accepted as a participant, not for CMT but early intervention for his GDD. Being accepted has been both a blessing and a curse. A blessing to have the funding to get the therapies he needs but a curse to have to fight to get funding for his CMT needs to enable him to participate to his best.

It has been very frustrating to have to explain to each new planner, therapist, teacher, agency, doctor and specialist about what CMT is.

To aid in this search I started seeking a local CMT support group to see if they could point me in the right direction. It took a few searches even to find the CMTAA head office in NSW. I contacted them and was informed to the best of their knowledge there were no services in my state, in fact they refer any enquires from my state to Melbourne for diagnosis and treatment. They did give me the contact details of the local coordinator in my state to speak with. This was the turning point for me as the coordinator was also a mother of someone with CMT and her input has been invaluable with my son.

I soon learned the invaluable input a support group has, to both carers and persons with the condition. A point of contact here to help support others with the condition, enhance their life and working to raise awareness of CMT. The CMTAA support enabled me to better develop the necessary skills to liaise with his preschool and school, to have things in place to allow him to attend mainstream school and participate to his ability whilst being supported when needed.

My biggest struggle at the moment is dealing with sourcing orthopaedic boots for my husband locally. Since moving here we have tried several places, all to no avail. We rely on getting his shoes made and sent over from Sydney. Unlike the locally made shoes that don’t work, the Sydney made boots he can wear without a problem. I’m now also sourcing shoes for my son suitable to wear with AFO, as off the shelf sneakers have to be so big to accommodate them as well as be able to do up. I have to source these from an American made shoe company or again pay huge money for custom made. Even just finding CMT aware professionals is a real headache and filling in documents to access services is nearly impossible due to the lack of government knowledge of the condition.
Support Groups
The CMTAA runs state and regional support groups in every state and territory of Australia. Groups offer the opportunity for pwCMT to communicate, share and learn with other people who also have CMT. Coordinators assist pwCMT and their families to source health care professionals who have experience with CMT patients. Regional CMTAA support groups offer a forum to discuss the challenges that individuals face. Evidence suggests that support and advocacy groups such as the CMTAA offer advantages ranging from empowerment and knowledge-sharing to psychological benefits to participants. Empirical and extensive meta-analysis studies confirm the efficacy of psychosocial intervention. Empirical studies of support groups analyzed randomized controlled trials, different professional support groups and numerous other studies to confirm the benefits of such organizations.

The CMTAA offers support for children with CMT. A youth camp is held annually for young people with CMT 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 managing CMT.

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

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

“It’s always awesome to meet people that go through the same challenges.”

Most children reported that they had not known someone with CMT of their age before, that they had been involved in numerous trials and felt like they “did not fit in” and that “no one else understood.” Many children who attended camps reported in their testimonials that they

86 http://psycnet.apa.org/journals/amp/55/2/205/.
88 Youth weekend proposal tendered to the CMTAA Executive Committee as part of application to run “Aussie Kids” a camp for children with CMT.
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.

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

Initially the camps were only held in NSW; however proposals have been made to hold camps across other parts of the nation in order to benefit a wider audience of youth with CMT.

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

Recurring words and themes in testimonials include ‘uplifting, confidence, interaction and belonging.’ Numerous studies have been conducted on the importance of support and advocacy groups; they have benefits ranging from empowerment and knowledge-sharing to psychological benefits to participants.

89 CMTAA Aussie Kids testimonials.
90 http://psycnet.apa.org/journals/amp/55/2/205/.
8. Education

Socialization and normative behaviors are learnt in the formative years of primary and secondary education and shape the rest of the life of an individual.⁹¹ Children with disability are more likely to stay in school longer, with a higher participation rate (82.1%) in secondary school than children without disability (77.0%).⁹² Secondary enrolment completion rates for the late onset group (75.8%) fall below the overall Australian disabled population frequency, remaining close to the rate of people without disability (77.0%). One explanation may be that many with late onset are not experiencing symptoms and/or having a diagnosis until after their secondary school years. The completion rate (87.9%) for early onset is higher than the Australian rate for all children with disabilities.

In every state, pwCMT completion rates outperform the national average of Australians with disability. Secondary completion for early onset pwCMT is highest in the ACT (92.0%) and lowest in NSW (84.2%). Among all Australian children with a disability, 65.9% attend mainstream schools and 9.9% attend special schools;⁹³ whereas children in the early onset group, just 3% attend a special school.

“My first school was the Illawarra Crippled Children’s School. My classmates had all sorts of conditions but most were severely mentally handicapped. My mother fought the NSW Dept. of Education until they let me into a standard state public school. It was 1972.”

Evidence suggests that special needs schools offer better outcomes for students, but children with CMT have until recent decades have been bundled into special needs schools, because mistakenly viewed as having a mental health issues. Although pwCMT are staying at school longer, SCWM data shows significant dissatisfaction at how well primary and secondary schools accommodated their CMT needs.

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⁹³ Ibid.
Early onset pwCMT show greater dissatisfaction with their schools’ ability to accommodate their needs compared to those with late onset. Male pwCMT reported higher dissatisfaction (53.4%) than female (41.4%).

Overall, most SCWM participants reported a high level of involvement in cultural and sporting activities, however the early onset group (11.69%) ‘never’ participated in cultural and sporting activities; twice the frequency as late onset (5.2%). Focus group data suggests that secondary schools struggled with students who had more severe core activity limitations. Students with CMT reported being restricted in their choice of subjects, with some reporting that they were offered alternative subjects and denied their first choice. Metalwork and woodwork were the
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subjects most frequently mentioned in focus groups as those denied for school children with CMT. Others reported being unable to enroll in home economics (cooking).

“I wanted to be a chef. When I put my name down to do cooking the teacher took me aside and explained that it was not safe for me or other students. That I might knock over a boiling pan…”

Denial of subject choice because of having CMT was one setback faced by children. Another setback faced by many was bullying, with half (51.7%) of SCWM participants reporting that they were bullied at school.

Data on bullying frequency in Australia is mixed (20% male and 15% female).\textsuperscript{94} However, even after sourcing scholarly data for the highest frequency available within the general Australian population (32%),\textsuperscript{95} children with CMT exhibited a higher frequency of being bullied at school. For 13.4% of SCWM participants, bullying was an event that occurred often. Bullied students are likely to suffer social withdrawal (73%), depression and decline in academic engagement and performance.\textsuperscript{96} Lack of academic engagement is of great concern for the future of young people with CMT in regard to employment options. Notwithstanding the challenges secondary schooling posed for children with CMT, as young adults they excel at the tertiary level.

Educational attainment profiles for female and male tertiary students outperform the general population at the certificate, diploma, bachelor degree and higher levels.

\textsuperscript{96} D., Shaw, T., Hearn, L., Epstein, M., Monks, H., Lester, L., & Thomas, L. 2009. Australian Covert Bullying Prevalence Study (ACBPS). Child Health Promotion Research Centre, Edith Cowan University, Perth.)
Analysis of the data based on sub-national jurisdictions indicates that each subset is performing at almost identical levels or better and producing postgraduate degree holders at above the general population frequency.

Educational attainment is an indicator of knowledge. Abilities obtained from the formal education process are crucial for young people with CMT. A progressively technological workforce requires employers to recruit employees with higher skill levels. Only individuals with secondary school qualifications have less chance of finding paid employment than those who have completed tertiary education. For people who have not achieved the HSC, the unemployment rate is 5.6% (2006). For those who have achieved the HSC as their highest level of education, the unemployment falls to 4.7% and unemployment for a bachelor degree or higher drops to just 2.2%.97

9. Employment and Income

Paid employment can provide pwCMT with confidence, increased societal understanding and social skills as well as prospects for career advancement by adding innovative work abilities and understanding. Progress towards achieving workplace equality and protection against discrimination for people with disabilities has been achieved since the Disability Discrimination Act 1992 (DDA). While some advancement in terms of ending discrimination has been made, people with disabilities are participating less in the labour force than the general Australian community.

Young people aged 20-29 with CMT are ten times more likely to draw the Disability Support Pension (DSP) than the same cohort in the general population. Across all age cohorts, pwCMT are far more likely to rely on the DSP as their main source of income.

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“I worked up until the fatigue that is caused by CMT made it too hard. I am on a pension now but I would prefer to be working.” Focus Group quotation.

Reliance on the DSP differs significantly between early and late onset CMT. Late onset DSP reliance peaks at 20.8% in the 40-44 range, whereas early onset peaks at the 25-29 range (17.1%) and at 10.1% under the age of 20.

For some focus group participants there were generational welfare concerns, but SCWM data shows that 18.7% of participants had already reached the age of retirement before diagnosis. For another 26.6% of pwCMT, diagnosis was within the last 5 years of their working lives, and CMT had little or no influence on the working lives of those aged in their sixth decade and onwards. Focus groups indicated that early retirement was even more frequent for people whose work involved an emphasis on strenuous physical labour.
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“Well, it [CMT] came on slowly. The driving was not the problem, it was more the loading the truck, walking up steep driveways without tripping and the fatigue. An office job was not on as I didn’t even finish school. I would have another 10 years of work in me if things were different” Focus Group quote.

Within the general population, labour force participation in Australia is at its highest within the 45-54 age cohort. However, labour force participation for people with disabilities is at its highest in the 25-34 age cohort and is at its lowest (40.9%) later in life (between 55-64 years of age). According to the ABS, the lower participation rate of people with disabilities in later years may partially be due to the “desire for retirement” or to “difficulties experienced by mature-age job seekers”.

“I would have loved to keep working but the CMT made it impossible. My mother also needed to stop working at around 35. Having kids, husband who works, and CMT, something had to give.” Focus Group quotation.

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Focus group data supported existing evidence that finds the disease is more detrimental to women remaining in the workforce in later life. Women with disabilities (49.0%) are less likely to participate in the labour force than men (56.6%). SCWM data also shows that both early and late onset pwCMT women were in full-time employment.

Early onset pwCMT earn less than late onset. In the $50,000 to $70,000 range, people with early onset (6.5%) earn less than half than do the late onset group (15.4%). Two-thirds of pwCMT between 18-64 years of age believe it unlikely that they will secure a suitable job in the next 6 months. It should be noted that while income data can be used to compare average earnings between early and late onset CMT, such comparisons do not take into account hours worked, which contribute significantly to the differences observed between early and late onset earnings.

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"I think the best chance to get a job is with the government. For sure I am going to keep trying to find work." Focus Group quotation.

Unfortunately the public sector at a federal level is not setting a good example for the private sector to follow. The number of people with disabilities employed by the Australian (commonwealth) government is in steady decline. People with disabilities made up only 3.8% of the Australian public service in 2002/2003, a decrease of 5.8% over a ten-year period.\textsuperscript{102}

SCWM data shows that two thirds (66.7%) of pwCMT believe employment agencies have a poor approach to assistance in seeking employment.

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“The best thing I ever did was get off Centrelink and start working. The extra money is great but I think the best part of it is that I like what I do.” Focus Group quote.

Some focus group participants had greater success with employment when using specialized employment agencies for people with disabilities. Despite the difficulties in finding work, once work is found most pwCMT enjoy their work (86%). This job satisfaction spills over to employment retention rates.

All things considered, how satisfied are you with the type of work that you do?

- Very satisfied: 44%
- Somewhat satisfied: 42%
- Somewhat dissatisfied: 11%
- Very dissatisfied: 3%

“It has been 23 years and I am still in the same job. I can’t imagine working some place else.” Focus Group quote.

Focus group discussion to do with paid employment indicated that paid work is needed in people’s lives. Focus group participants showed a strong desire to contribute to the economy and to society generally. A common theme was that not only did a job provide income but that it gave pwCMT a greater sense of purpose and a way in which they can contribute to society. A body of scholarly work indicates that paid employment is closely linked with mental health.103

For some, however, the challenges of having CMT restricted their employment options. The greater the core activity limitation severity, the less likelihood there is of a person’s ability to participate in the labour force. Those with mild or moderate limitations in the 15-64 age cohort participate at a rate of 52.5%. However, among people with severe or profound limitations, the


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labour force participation rate (29.7%) drops substantially. This pattern was evident across all types of disability. For example, the participation rate of those with moderate or mild physical restriction was 48.8%, while the participation rate of those with profound or severe physical restriction was 26.3%.

CMT is a physical disability and people with physical disability have a significantly lower unemployment rate (8.1%) than those with even a moderate to mild intellectual disability (24.7%). PwCMT are more likely to participate in the labour force than people with intellectual disabilities who face different barriers in training and education. Typically, for any person with a disability in the workforce, the more severe the core activity limitation, the fewer hours they are able to work. This is reflected in part-time participation frequency. Individuals with a profound or severe core activity limitation who worked were more likely to work part-time.

“In 2012, there were 2.2 million (14.4%) Australians aged 15-64 years, of 'prime working age', with disability.”104

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SCWM data for early onset pwCMT labour force participation is in line with SDAC findings, in that disability frequency typically increases with age. The disability frequency for those who are 15-24 years of age (7.9%) increases with age (18.0% for 45-54 and 29.0% for people 55-64). As late onset disability can occur from the second decade of life to the age of retirement or after, labour force frequency is likely to mirror the general population until symptoms present and diagnosis occurs. However, the early onset group differs fundamentally from the late onset group, with symptoms occurring within the first decade of life. Both early and late onset live a normal life span,105 and thus the labour force loss is typically the duration of the entire ‘working live’ of a pwCMT. To measure the precise impact of this highly variable disease on labour force participation requires further longitudinal research. Nevertheless, labour force participation observations in this study can gauge the number and proportion of pwCMT who are actively looking for work or are employed.

In the post DDA period, labour force participation rates for people with disability 15-64 years of age have remained constant (54.9% to 52.8%). However, the participation rate for 15-64 year olds with no disability increased from 76.9% to 82.5% in the same period. Unemployment for 15-64 year olds with a disability has declined in the last two decades (17.8% to 9.4%), as also for people without disability (12.0% to 4.9%). Nevertheless unemployment rates for people with disability continue to be significantly higher than for those without disability.106

Of all 15-64 year old people with a disability in Australia, nearly half (47.3%) were looking for work or in employment, and 33.6% in this group were permanently unable to work. The early onset cohort found it more difficult when looking for work (64.8%) than late onset (24.4%). However, 19.3% reported that their disability did not preventing them from working, and they are thus considered to have no employment restriction.107

Of the states and territories, the ACT has the highest labour force participation rate (66.6%) for pwCMT. The ACT also has the best performing participation rate for all people with any disability. Tasmania has the lowest participation rate of all people with disabilities, while for pwCMT Queensland has the lowest rate (26.8%). More men than women participated in the labour force in each state, except South Australia, which was the only state or territory with

107 Ibid.
more women than men with CMT in the labour force. The ACT had the lowest unemployment rate (4.9%), and Tasmania had the highest (14.9%), compared to people without disability, and NT had the lowest rate with 2.6% and Tasmania had the highest at 5.3%.108

While pwCMT follow a similar trend to Australians with a disability in part-time employment level, this is not the case with underemployment. Approximately one in three Australians with a disability (32.4%) who work part-time prefer to work more hours, as opposed to 27.1% of people without a disability. SCWM data indicates a lower level of underemployment for pwCMT, with just 11.5% who would choose to work more hours.

Underemployment changes with the severity of the core activity limitation. Early onset pwCMT show a higher frequency of underemployment (39.7%) compared to late onset (12.6%). In a non-material sense, the rewarding nature of work had much to do with pwCMT maintaining their paid employment. Focus group participants highlighted the ability to balance their work life with CMT-related health commitments. Often this meant reducing the number of hours worked away from full-time to part-time or casual work.

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“Human resources were more than happy to accommodate my CMT needs. Dropping my hours meant I could keep working.” Focus Group quote.

The most frequent reason given for working less than a full-time workload was having CMT (30.8%). In 27.8% of cases, pwCMT needed to change their occupation altogether.

Has having CMT ever caused you to change occupation?

- Yes 28%
- No 68%
- Not sure 4%
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Changing occupation may not necessarily mean leaving the existing employer or industry. Many people reported that they were able to move away from labour-intensive work within the same organisation to better accommodate their individual working CMT restrictions. Focus group data was polarised to do with people’s perceptions on how worsening core activity limitation due to CMT affected their employment status. PwCMT who had diverse skills and/or tertiary education proved to be better positioned.

SCWM data shows that across both early and late onset pwCMT, 38.3% believed it very unlikely that they would have sufficient superannuation in place for retirement. The early onset pwCMT group expressed greater concern about having enough superannuation, with 61.4% believing it very unlikely that they will have sufficient.

“It is a double edged sword. Having CMT stopped me working 10 years before my time. And having CMT means I’m going to be less independent in my old age and need extra money because of that.” Focus Group quote.
10. Recommendations from this report

There was a vast amount of primary data amassed for this study. The two hundred question strong SCWM was completed by 418 individuals with CMT. Twenty-one focus groups were held in every state with 178 participants comprising of pwCMT, family, carers and partners. It is envisaged that this data collected will take the author years to interpret with many journal papers already planned. The Recommendations to address the Department of Health’s project criteria are as follows:

Major recommendation

1.1 Establishment of a national adult and paediatric CMT multidisciplinary clinic (the clinic).

1.1.1 Patient-centric multi-disciplinary CMT clinic to be operated by CMT specific clinicians. Primary and secondary care by a team of health care professionals who have expertise in CMT.

1.1.2 Clinic should be located in an Australian capital city.

1.1.3 The clinic to assess patients periodically with a focus on communication between the clinic and health care professional located in the patient’s residential area.

1.1.4 While the clinic should operate primarily with a patient-centric focus it should also contribute to CMT research.

1.1.5 Clinic should establish and maintain a register of people with CMT who are willing to participate for clinical trials and candidate therapies.

1.1.6 The clinic to formally correspond with international counterparts and the Inherited Neuropathies Consortium.109

1.1.7 Where possible, the following areas should be offered either at the clinic or by separate external referral.

a. Neurology/Physiatry

b. Orthopaedic Surgery

c. Physical Therapy

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109 The Inherited Neuropathies Consortium is an integrated group of academic medical centres, patient support organizations, and clinical research resources dedicated to conducting clinical research in different forms of CMT and improving the care of patients. Funded by the National Institutes of Health, the INC is part of the Rare Diseases Clinical Research Network.
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d. Podiatry

e. Orthotics/Prosthetics

f. Durable Medical Equipment

g. Occupational Therapy

h. Support group (CMTAA)

i. Genetic Counseling

j. Social Work/Counseling

k. Psychiatry

l. Nutrition

Key recommendations

2.1 A Department of Health initiative to improve community and medical awareness to do with genetic discrimination. To seek a comprehensive clinical genetics sector commitment and for regulating, research and Commonwealth Government policy development to enhance genetic testing technology to its full potential.

2.1.1 This study endorses the Australian Law Reform Commission’s key recommendation that; “Discrimination laws should be amended to prohibit discrimination based on a person’s real or perceived genetic status.”

3.1 The Department of Health provide significant and direct funding to the CMTAA. In recent years the CMTAA has been called upon to operating beyond its charter (support and advocacy) to assist investigators to harvest participants for vital research. Further, the CMTAA disseminates vital information to people with CMT that has a direct relationship in reducing the burden on the health system.

4.1 The Department of Health to initiate and fund an awareness campaign in collaboration with the CMTAA designed to educate the public of this most common inherited neurological disease. It is recommended that advertising posters and/or information broachers be produced to display in offices of General Practitioners and health care professionals.

Appendices

Appendix 1. How would you rate your overall physical health?

Appendix 2. Please rate the following aspects of your health:

Appendix 3. Co-existing conditions

Appendix 4. Signs and symptoms

Appendix 5. Number of times fallen in the past 4 weeks

Appendix 6. Broken or fractured bone resulting from a fall

Appendix 7. How would you rate your overall mental health?

Appendix 8. Do you feel that you have ever experienced reoccurring episodes of anxiety?

Appendix 9. Do you ever feel anxious about leaving the house?
Appendix 1

How would you rate your overall physical health?

- Very good: 7.6%
- Good: 16.2%
- Fair: 27.2%
- Poor: 7.3%
- Very Poor: 41.8%

Appendix 2

Please rate the following aspects of your health:

- Very poor
- Poor
- Fair
- Good
- Very good

Long term memory?
Short term memory?
Level of fitness?
Hearing?
Eyesight?
Teeth and gums?
Reducing the Health Burden of Charcot-Marie-Tooth in Australia

Appendix 3

### Co-existing conditions

- Migraine headaches: 11.9%
- Liver disease: 2.4%
- Kidney disease: 1.5%
- Cancer (any type): 12.8%
- Circulatory condition (e.g., stroke, hardening of arteries): 7.6%
- Obesity: 10.3%
- Type 2 Diabetes: 5.8%
- Type 1 Diabetes: 3.3%
- Any other mental illness: 2.1%
- Anxiety: 25.5%
- Depression: 24.9%
- Emphysema: 18.2%
- Tinitus: 17.0%
- Sleep Apnoea: 16.1%
- Asthma: 17.9%
- Scoliosis: 8.8%
- Heart disease: 10.3%
- Low blood pressure (hypotension): 37.4%
- High blood pressure (hypertension): 49.2%
- Arthritis or Osteoporosis: 0%

### Signs and symptoms

- Burns or scalds: 73.9%
- Trips or falls: 44.8%
- Difficulty moving around in the dark: 70.9%
- Difficulty learning or understanding things: 12.5%
- Difficulty gripping things: 7.2%
- Speech problems: 9.9%
- Impotence: 22.7%
- Erratic eye movement (squint): 20.0%
- Shortness of breath or difficulty breathing: 29.1%
- Incontinence: 3.7%
- Impaired ability to regulate body temperature: 82.1%
- Blackouts, fainting or loss of consciousness: 93.9%
- Weakness in feet and legs: 74.1%
- Weakness in arms and hands: 53.9%
- Loss of sensation in feet and legs: 0.0%
- Loss of sensation in arms and hands: 100.0%
Appendix 4.

Number of times fallen in the past 4 weeks

- 0: 3.4%
- 1: 16.9%
- 2: 19.8%
- 3: 3.4%
- 4: 4.2%
- 5 or more: 52.2%

Appendix 5.

Broken or fractured bone resulting from a fall

- Yes: 39.3%
- No: 60.7%

Appendix 6.

How would you rate your overall mental health?

- Very good: 1.6%
- Good: 6.1%
- Fair: 18.3%
- Poor: 32.9%
- Very Poor: 41.1%
Appendix 7.

Do you feel that you have ever experienced reoccurring episodes of anxiety?

- Yes: 12.7%
- No: 37.6%
- Not sure: 49.7%

Appendix 8.

Do you ever feel anxious about leaving the house?

- Yes: 3.7%
- No: 23.3%
- Not sure: 73.0%