Summary of research findings

Understanding how people are affected by Charcot-Marie-Tooth Disease (CMT) in New Zealand

Tēnā rawa atu koe, fa'afetai and thank you so much for taking part in the ImpactCMT study. We greatly appreciate you sharing your experiences of CMT with us.

This is an update about the findings from this research and our plans for communicating these outcomes. This information will be shared with individuals and organisations involved in the health, disability and scientific communities, both here in New Zealand and the rest of the world.

What we aimed to do

The Impact CMT study aimed to find out how many people are living with CMT in New Zealand and to help us understand how the condition affects peoples' lives. Previously information about how many people are affected by CMT was only available from studies conducted overseas.

What we did

This research study was funded by Neuromuscular Research New Zealand and was conducted in partnership with the Muscular Dystrophy Association, the New Zealand Neuromuscular Disease Registry and Auckland District Health Board (DHB).

A team of researchers and clinicians got together to try and find every living person in the Auckland, Counties Manukau and Waitemata DHBs. The greater Auckland region was chosen because it has neurogenetic clinics and a centralised neurology service and because these three DHBs cover a substantial proportion of the country's population.

This meant we could then determine more confidently how many people are living in the whole country with CMT. Just like the national census which counts everyone on a particular date, we chose 1st June 2016 as our "census" day.

When a person was identified, we offered everyone we could contact a chance to complete an interview about how the condition affected their lives.

What we found

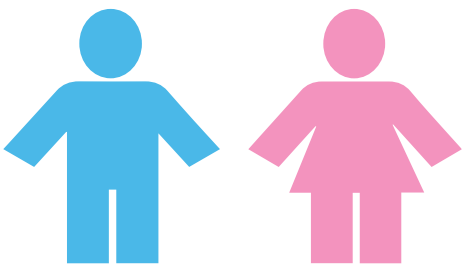
We identified 237 people with a diagnosis of CMT living in Auckland. From this, we estimate that there are about 710 people living with CMT in New Zealand. The frequency of CMT here in New Zealand was slightly lower than found in other countries.

There was an even gender split with half of the sample (50%) being male.

As expected, most people had CMT type 1 where the myelin surrounding the nerve is affected causing a peripheral neuropathy characterized by distal muscle weakness and atrophy, sensory loss, and slow nerve conduction velocity. More than 80% had CMT1A, which is caused by a duplication of the PMP22 gene.

76% of people had received a genetic test, with two thirds of tests providing molecular confirmation of diagnosis.

Of the estimated 710 people with CMT in NZ half are male and half are female.

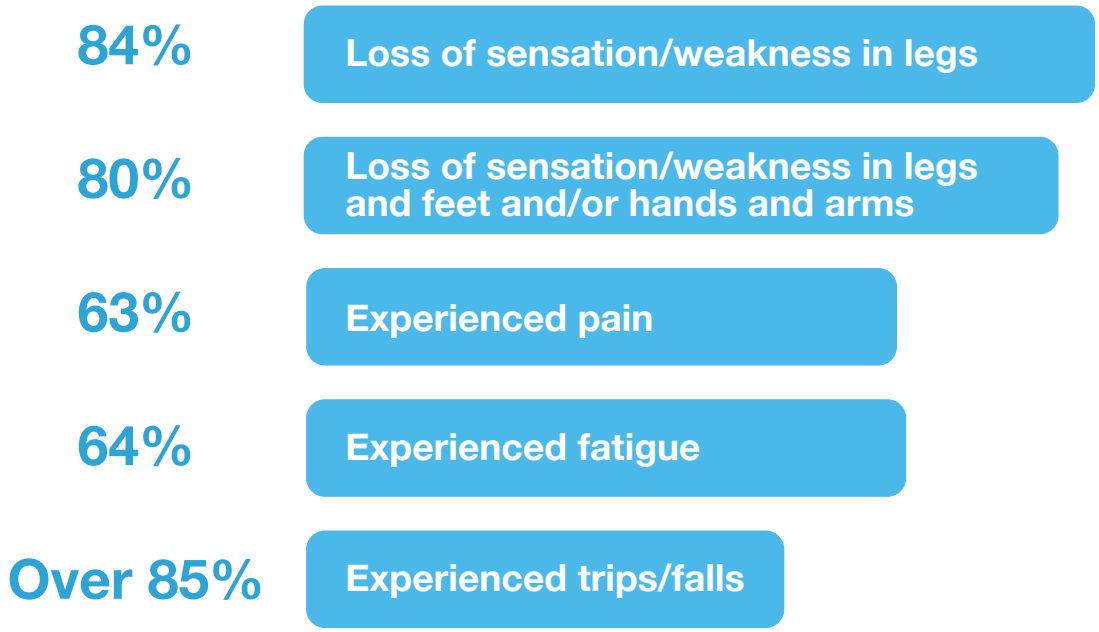


The proportion (12%) of children identified is lower than overseas estimates. This might be because we do not routinely offer pre-symptomatic testing for CMT in NZ as they do in other countries. We found that the frequency of CMT remains stable across age groups, meaning that CMT rarely shortens life.

More than half of people interviewed had at last one other family member who was also affected by CMT.

Symptoms

The most common symptoms for adults and children diagnosed with CMT were loss of sensation and weakness in arms and legs. More than half of people experienced pain, fatigue and trips/falls.



Levels of fatigue and muscle weakness were found to be linked to how well people were able to complete everyday tasks such as putting on a t-shirt or walking round the block.

Other conditions

Just under half (46%) of people also had at least one other medical condition which made it difficult to know what was related to the CMT and what was due to another condition.

Age of diagnosis

On average, people received a diagnosis of CMT in their mid 30s (mean age 35 years).

Satisfaction with health care

Most people were satisfied with the level of health care that they received with an average rating of 7 out of 10.

People commonly said that they wanted more information about CMT to be made available so they themselves, friends and family and health professionals would be more knowledgeable. People also said that they wanted to know more about how things might change for them over time and how to prevent further complications. It was also felt that follow up appointments with neurologists would be more useful if they were scheduled as the condition changed, rather than being at fixed intervals which were too far apart.

Just under a third of participants stated that they had at least one unmet need. Common unmet needs were not having a special mattress, equipment (such as hand rails) in the bathroom and aids to support communication (such as a hearing aid).

Overall, we found that people were functioning relatively well in everyday life. There were a wide range of experiences from some reporting minor impacts on their life whilst others were finding it difficult to manage. People said that whilst it was important to receive support to manage the physical challenges they encountered, the psychological impact of the condition was often neglected. People wanted more support in dealing with changing circumstances and living with an uncertain future.

What happens next

The findings of this research have improved our understanding of CMT in New Zealand. We will be writing up the results for an academic journal and will present them at conferences here in New Zealand and overseas. We will be discussing the findings with health professionals and service providers and hope they will help to increase the support available to adults and children living with this condition.

Thank you again for your valuable contributions to this study. For any comments/queries about the ImpactCMT study please contact Alice Theadom on email: alice.theadom@aut.ac.nz or 0212460728.