

MD-PREV

PREVALENCE AND IMPACT
OF GENETIC MUSCLE
DISORDERS IN
NEW ZEALAND



Summary of research findings

Tēnē rawa atu koe, fa'afetai and thank you so much for taking part in the MD-Prev study. We were honoured to hear people's stories and greatly appreciated you sharing your experience of living with a genetic muscle disorder with us.

This is an update about the initial 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 did the MD-Prev study aim to do?

The MD-Prev study started in 2014 and aimed to find out how many people were living with a genetic muscle disorder in New Zealand on 1st April 2015. We also wanted to find out how these conditions affected individuals and their family/whānau's lives.



How many people are living with a genetic muscle disorder in New Zealand?

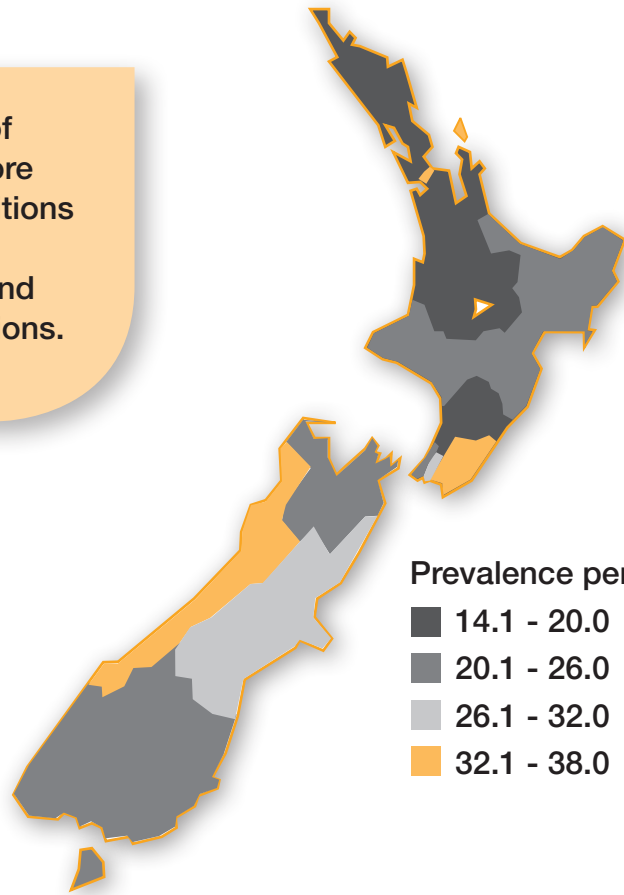
For the conditions that were included in the study, we identified 966 people with a clinical and/or a genetic diagnosis of a genetic muscle disorder. This included 159 children (under 16 years of age) and 807 adults.

Apart from the conditions which only occur in males or females (e.g. Duchenne muscular dystrophy or manifesting carriers) there was an even split of gender for the other conditions.

This table shows how many people had each of the conditions included in the study

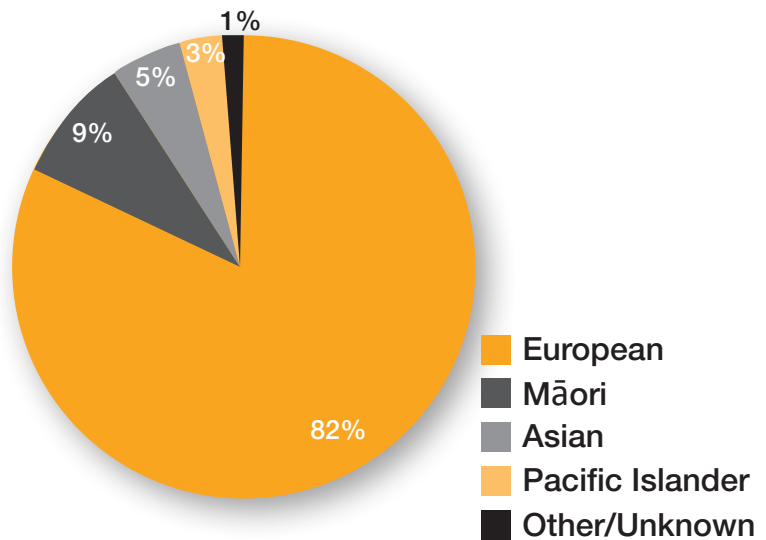
Condition	Total number of cases in NZ	Percentage of total with each condition
Duchenne muscular dystrophy	104	10.8
Becker muscular dystrophy	71	7.3
Manifesting carrier of dystrophinopathy	15	1.6
Myotonic dystrophy	343	35.5
Facioscapulohumeral muscular dystrophy	123	12.7
Emery-Dreifuss muscular dystrophy	11	1.1
Limb girdle muscular dystrophy	93	9.6
Congenital muscular dystrophy	27	2.8
Distal muscular dystrophy	9	0.9
Congenital myopathy	60	6.2
Other myopathies	10	1
Ion channel muscle diseases	66	6.8
Pompe disease	10	1
Unspecified	24	2.5
Total	966	100

Taking into account the size of the population, there were more people living with these conditions in the Wairarapa, West Coast, South Canterbury and Auckland City District Health Board regions.



One in ten people living with a genetic muscle disorder were not known to a health care or community service.

Study participants identified with a range of ethnicities. There were fewer Māori and Pasifika people than we were expecting based on representation of these ethnicities in the general population.



What is it like living with a genetic muscle disorder?

We interviewed 581 people to further understand the experience of living with a genetic muscle disorder. We found that the experience varied a lot, even when we looked at people with the same diagnosis.

Symptoms

The most common symptoms across all conditions were muscle weakness, poor balance and limited movement. More than half of those interviewed described experiencing pain and fatigue (extreme daytime tiredness). Other common difficulties included visual disturbance, muscle spasms and headaches.

Whilst the physical impact of the condition was important, people felt that the psychological impact (such as managing uncertainty about the future) was often not recognised.

For example, one person said;

“The psychological impact is harder to deal with than the physical. I do wonder about what my future holds, especially as I age.”

Information on care and satisfaction

Overall more than 85% of adults, and parents of children living with a genetic muscle disorder, were happy with the health care that they had received. However, more than half of people wanted more information about their condition, genetic counselling, information about signs and symptoms to look out for, what might happen over time and how to prevent further complications.



More general awareness of these conditions was also felt to be needed by GPs and those in the community to enable people to understand and assist those living with these conditions better.

People also wanted to see their neurologist more frequently and for it be easier to access medical support as their condition changed.

For example, one person said;

“I see my neurologist every 2-3 years, yearly would be better as there's no other contact with anyone. GPs are limited in their knowledge and change often.”

Moving around in the community

One in twelve adults and one in ten children reported that they needed ramps (both portable and installed) to facilitate access to their home but either did not have these or needed additional ones. It was identified that access to other people's homes was particularly challenging making it more difficult to socialise with others. Bus and railway stations were also quite difficult to use.

1 in 12 adults and 1 in 10 children needed ramps



When services were available people felt that they had to 'fit in with the service' rather than services fitting their needs. This impacted the way people lived their lives, particularly in relation to personal supports. For example one person said;

“ I have no choice about what time I go to bed and get up, because I'm dependent on carers. Naturally I'm a night owl but I have to be put to bed at 8.30pm. By the time my morning carers arrive to get me up I'm desperate to go to the toilet.”

Impact on relationships

Just over half of adults felt that their condition impacted on their romantic relationships. In some cases this brought people closer together. In other cases, the challenges of living with a genetic muscle disorder placed a strain on relationships or they became difficult to maintain. For example, many found trying to manage their fatigue or breathing difficulties made it difficult to do activities together or to have the energy to put into the relationship. This was also true of parents of a child with a condition.

How will information from the study be used?

We hope that the findings of this research will improve our understanding of these conditions and increase the support available to people living with a genetic muscle disorder.

We are talking with clinicians, health policy makers and service providers about the results, particularly around access to services, resources and providing more information to people, at different stages and as their needs change. We are also discussing the findings about the difficulties people experienced accessing the community with disability advocacy groups.

The findings will also be used in the training of health professionals, such as nursing students, psychologists and physiotherapists, about working with people with genetic muscle disorders. We will also write up the results for academic journals and present them at conferences here in New Zealand and overseas.

How will my privacy be protected?

When we share information about this study, it will not contain any material that could identify you. We have removed any specific information relating to you (e.g. names, addresses and date of birth). Information will only be grouped and presented by diagnosis, age, gender, ethnicity or geographic location.

What do I do if I want to take part in other research studies?



The New Zealand Neuromuscular Disease Registry helps to connect researchers with people with neuromuscular conditions. We have enclosed an information sheet about the Registry. You can contact Miriam Rodrigues on email: registry@mda.org.nz or call/text 027 468 8044 if you are interested in finding out more.

What do I do if I need further support?



**Muscular Dystrophy
New Zealand**

The Muscular Dystrophy Association of New Zealand (MDANZ) offers information, support and resources to people living with all of the conditions included in this study. We have enclosed a leaflet about their Fieldwork service. If you would like to talk with someone about this and the other services and benefits provided by MDANZ, please contact them on 0800 800 337, or email info@mda.org.nz

What do I do if I have questions about this research?

If you want more information about this research please feel free to contact the lead researcher Alice Theadom on email alice.theadom@aut.ac.nz or phone 09 921 9999 extension 7805

We thank all of our supporters who have helped make this study happen including Genetic Health Services NZ, Ministry of Health, Muscular Dystrophy New Zealand and our funders the Brendel Trust and the Health Research Council.