InTouch

KIA NOHO TATA Inclusive | Inspiring | Informative Spring 2017 Issue 96

We are family

Three generations, one condition

Impact CMT

NZ study results explained

Freedom month

Join our campaign

Time out

The benefits of relaxing

Muscular Dystrophy New Zealand

InTouch

PO Box 12063, Penrose,
Auckland 1642, New Zealand.
Freephone 0800 800 337
NZ Phone: (09) 815 0247
International prefix (00649)
Fax: (09) 815 7260

Editor: MDANZ National Office
media@mda.org.nz
0800 800 337

Design: The Artset
the.artset@gmail.com

Cover photography: Gemma Foulds
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Also thanks to Allied Medical, Biogen and Sanofi Genzyme, the ARA Lodge No 348 IC Charitable Trust, the Clyde Graham Trust, NZ Post Community Post, Auckland Council, Richdale Charitable Trust and the Independent Living Service for their continuing support.

Korero from Ken

Ngā mihi nui ki a koe arā me tō whānau hoki.
Greetings to you and your family also.

Our April AGM in Hamilton was a mixed blessing. Firstly, it was sad to see Heather our past chairperson and Sophie, vice chairperson, standing down after distinguished and dedicated service to MDANZ. My sincere thanks to them both. However, I am delighted to welcome the new members onto our National Council. We have a very strong and dedicated team.

I am humbled that members have placed their trust in me to lead the organisation. I pledge to use my business experience and finance skills to help it become even stronger and more sustainable. My primary focus is making a difference in the lives of our members.

For the first time in MDANZ's nearly 60-year history, both the Chief Executive, and myself as Chairperson of National Council, have firsthand experience of what it is like to live with, and face the challenges of a neuromuscular condition. Having an organisation led by people with lived experience brings a unique perspective and strength, because we know the difference the support of a professional, caring and well-resourced organisation brings.

As winter has taken hold, I remind you one of the important benefits MDANZ offers members is reimbursement for the pneumonia vaccine. Having taken advice from my GP, I got the vaccine and urge all members to consider it.

Looking ahead to our appeal month, September also has a special day to celebrate fathers, so I would like to take this opportunity to think of our parents and the roles they have played in our lives.

Ma te Atua koe e manaāki e tiaki
May God bless and keep you

Ken

Ken Green
MDANZ Chairperson

It might be you ...

or a family member, a neighbour or a friend. It could be a wee baby, or a retiree, it could happen at any stage in life.

Muscle weakness and wasting conditions can strike anyone of any age, of any ethnicity. These disabling conditions are called neuromuscular conditions with most but not all being genetic in origin.

We provide services to people with neuromuscular conditions – services that are unique and help them to live their life to its fullest.

You can help by:

- **Telling family members affected by a neuromuscular condition about us.**
- **Supporting our fundraising efforts.**

Muscular Dystrophy Association Patron, Judy Bailey.

In touch with Ronelle

Freedom means different things to different people.
This September we ask what it means to you.

Whether it's Janis Joplin or Pharrell Williams, using the word freedom in a lyric can make an ordinary song, an enduring classic. We think our vision *Freedom Beyond Limits* will also stand the test of time.

For our annual appeal this year, we are building on the Freedom relaunch which took place in 2016. In recognition of global awareness days for several of the conditions that MDANZ covers, this September we will be holding a month-long Freedom campaign.

Last year, we were really impressed at how our members and supporters got involved in our conversation about Freedom. We want to keep the conversation going for another year and ask, "What does Freedom mean to you?"

Let's join voices and raise awareness about our needs and aspirations. In my experience, it is the everyday freedoms that can easily be taken for granted. They include driving my own vehicle, independently cooking a meal, having a career and being a parent. Your idea of Freedom might be similar or different – and we want to hear about that.

Join us this September to raise awareness and vital funds for the thousands of Kiwis affected by neuromuscular conditions.

There are many ways you can get involved, whether it be through social media, volunteering to fundraise for your local branch, displaying a collection box in your workplace or a local business, sharing a picture or video talking about what freedom means to you, or holding a freedom party or mufti day at school.

Let's be loud and proud and educate our society this September. Be free, be yourself.

Ngā mihi mahana,
Warm regards

Ronelle Baker
Chief Executive

It was great to have all the team together at National Office for our regular staff development and training. As it was right in the middle of winter, we decided to be a bit festive and held a Wicked Secret Santa session, where everyone bought an unwanted present to re-gift to another member of the team.

A cup of tea and a catch up with … Gemma Foulds

Each issue we introduce a MDANZ team member:

How long have you worked for the Muscular Dystrophy Association and what do you do?

I'm the Office Manager for the Canterbury Branch, I am new to the role and am very excited to make a difference to the community here. I will be helping Southern Branch too.

What qualifies as a great day at work for you?

For me, a great day means making sure our members are happy, and working as a team. Somewhere in there, I would include completing my to-do list.

If resources and funds weren't an issue, what would you like to see our members enjoying?

I would like to see our members have greater access to medical care and medication, transport and equipment, so that they are able to have the best possible life, and the freedom to be able to make the right choices for themselves and their families. These things add to the well-being of the individual. Freedom is often taken for granted and can be lost easily.

What's the perfect morning tea for an office shout?

There's so much to choose from, but I would nail it down to the good ol' classic lamingtons, cheese scrolls, sausage rolls and lots of coffee. Nothing beats getting together to share stories and having a laugh.

What are you passionate about?

Photography is my biggest passion. Over the past seven years, my hobby has turned into a business. My passion now centres around creating powerful images for non- profit organisations and taking striking images around chronic health, terminal illness and disabilities. I want to give our community a greater awareness of these conditions by showing a different perspective to how they are often viewed. I believe in giving back and advocate for equality in the workplace and our community at large. Let's help each other to do great things.

Thanks to Gemma for taking our cover image for this issue.

Learning to lead

Latifa Daud tells us about being part of the Be.Leadership programme

When I applied last year, I had absolutely no idea how life-changing and invigorating the programme would be.

We are just over half-way through the year-long programme, and my thoughts and ideas have been challenged and strengthened. I am excited to see how much more I can grow in the second half of the programme.

As far as leadership goes, our group has been encouraged to think beyond our traditional definitions of the word.

Our world is changing every day, which presents an exciting challenge for people within the access community to become a core element to this change. I think it is incredibly important for us to rise to this challenge and harness the leadership potential we all have within us.

While the traditional model of disability puts us in a position of weakness, this programme encourages us to be in the world in a way that flips this weakness on its head. It encourages us to think about how we can challenge the status quo and seek greater inclusion for communities that are currently marginalised by society.

Latifa Daud

Applications for the Be.Leadership Programme 2018 are open until 31st October. Find out more at www.beaccessible.org.nz

Our first Dukies

Adventure awaits

Our Award Unit Leader and fieldworker based in Nelson, Marty Price, is delighted to share the news that five of our young members have signed up to begin the Bronze level of the Duke of Edinburgh's International Award this year! "This is such an amazing opportunity for our young people to set goals and stretch out of their comfort zones and be recognised for their achievements with the full support of MDANZ behind them".

Our participants are spread out throughout the country.

Grace from New Plymouth is looking forward to building on what she knows, as well as gaining some new skills.

Ciaran from Nelson is excited about making new friends, while gaining work experience and getting motivated for his future.

Ella from Waiuku has already set her goals for what she wants to achieve, including helping in a cat colony and playing badminton.

Jack from Invercargill is already doing great things, training for the New York Marathon.

Dylan from Auckland is hoping to learn new skills and experience new things.

THE DUKE OF EDINBURGH'S INTERNATIONAL AWARD
AOTEAROA NEW ZEALAND | HILLARY AWARD

MDA news

Members' Discretionary Fund

Sharing vital funds

We are halfway through our second year of the members' discretionary fund. With two funding rounds already completed, we have contributed around $12,000 to our members. While we do not have the funds to assist everyone who applies, we have been able to offer 25 members some vital funds towards products, services or experiences that are life-changing.

These have included:

- Contributions towards travel overseas to attend conferences and participate in sporting events
- Funding towards the purchase of aids for hearing and mobility
- Contributions towards educational programmes

For an application form and more information about the fund, including guidelines, please go to our website, www.mda.org.nz.

Jenny Smyth at football practice. She received funds toward a new set of motors for her sports power chair.

Support Us

Any donation, big or small makes a difference. Donations of $5 or more are tax deductible.

Call: 0900 426 93 to make an automatic $15 donation.

Online: Donate any amount securely online.
www.mda.org.nz

Bubble soccer

Having fun while fundraising

An enormous thank you goes to Georgia Mayor, a student at Onewhero Area School, and others in her business class who ran a Bubble Soccer tournament at school and at the Onewhero Rugby Club as part of their Level 2 Business assessment. They held a child and adult tournament with prizes and food, which was open to all members of the local community.

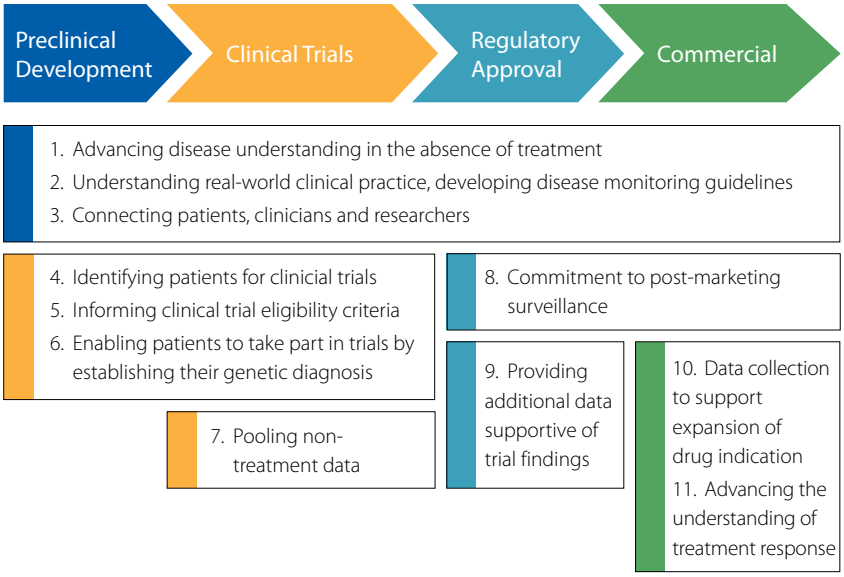
The students decided to donate the proceeds of the event to MDANZ, inspired by friends they have who live with muscular dystrophy.

Chief Executive Ronelle Baker spoke at a school assembly to thank the students and present them with certificates to thank them for their amazing generosity.

Bubble soccer lets you play inside an inflatable suit. Thanks to the students who organised the event.

The development of the registry

Our five-year evolution



The New Zealand Neuromuscular Disease (NZ NMD) Registry has recently published a report in the Journal of Neuromuscular Diseases introducing a model of registry development. A registry's role is dynamic and should be responsive to the changing needs of its stakeholders; people with the condition, researchers and clinicians as illustrated in the Bogard model of registry development (see diagram).

In five years the NZ NMD Registry has evolved from carrying out roles important during preclinical drug development, shown as steps 1-3 in the model, to performing vital work in the clinical trial arena by identifying patients for clinical trials, and Informing study eligibility criteria (steps 4 & 5).

As treatments become available, post-marketing surveillance (step 8) becomes a consideration and this is now relevant for both Duchenne muscular dystrophy (DMD) and spinal muscular atrophy (SMA). The registry will continue to playing a role, performing steps 8-11 as more treatments enter the commercial market.

NZNMD REGISTRY

Trustee changes

Farewell and thank you to Dr Sophie Tauwehe Tamati. We welcome Dr Tristram Ingham as a newly elected Councillor for MDANZ and the newly appointed representative to the NRFT.

Investigating the diagnosis experience

Neuromuscular Research New Zealand post-graduate grant recipient Miriam Rodrigues, has begun her PhD at the University of Auckland's School of Medicine. Miriam is investigating people's experience of receiving a genetic diagnosis and also their attitudes towards preconception screening for genetic muscle disorders. This research is a sub-study of the MD Prev study and Miriam will be specifically analysing some of the responses to the impact assessment that was delivered as part of that larger study.

She is interested in identifying any factors that have resulted in a better experience of diagnosis for both adults receiving a diagnosis, and for parents of children being diagnosed. Miriam will be looking across a range of different genetic muscle conditions, which have varying inheritance patterns and severity. When not working on her PhD, she is the curator of the NZ NMD Registry and also advises on the programmes and services MDANZ offers.

Jack proudly wearing his Freedom T shirt with his Mum and number one supporter.

My Freedom

Taking on the Big Apple.

If you're looking for someone who is living the MDANZ vision of *Freedom Beyond Limits*, you don't need to look any further than Jack Lovett-Hurst.

This November, Jack is heading to the USA to compete in the New York Marathon on his hand cycle. Jack's mum and step dad, Debbie and Greg Houkamau, are his number one supporters and trainers and will be going along with him.

Jack is training at least four times a week, and while it's hard work, it gives him a glimpse of what's ahead.

"Every time I train I get really excited,' he says.

Let's talk about Freedom

We want to know what it means to you.

Last year we relaunched our annual fundraising and awareness campaign with a Week of Freedom. This year we are going even bigger with a Month of Freedom, building on last year's success, and the 2016 launch of our new strategic plan which engages our community in a conversation about Freedom. The 2017 Freedom campaign will be launched to coincide with a global awareness campaign, Duchenne Awareness Day on 7th September.

We want to start a conversation amongst our members, and all New Zealanders, asking, "What does Freedom mean to you?"

For our members these may well centre on everyday freedoms that can easily be taken for granted, like driving their own vehicle, living independently in their own home, making a cup of tea, cooking a meal, to broader concepts like having a career and relationships, or parenthood.

Get involved

We need your help to make our campaign a success.

There are many ways you can get behind MDANZ during the Month of Freedom;

- Volunteer to fundraise for your local branch
- Display one of our donation boxes in your workplace
- Like us on Facebook
- Share a picture or video talking about what Freedom means to you
- Challenge your friends and family to go without something they can't live easily without – like electrical appliances, their car, or mobile phone for a day – if they can't they need to make a donation to your local branch or MDANZ.
- Hold a Freedom party
- Buy a unique gift from MDANZ – we have limited edition Freedom merchandise for sale.

Making Freedom permanent

Miriam Hanna is getting a tattoo to make a permanent statement.

Miriam Hanna with daughter Christa.

It's coming up to a year since I joined the Muscular Dystrophy Association of New Zealand as Information and Resource Coordinator. I started just before the September campaign last year, and this year am very excited to be part of the planning and organising at National Office. I wanted to do something personally that could help raise funds for the great work the organisation does, and for the beautiful people who are our members. I have been thinking about getting a tattoo for a while, and really like the idea of making a permanent statement about the value I place on freedom.

The design of the tattoo I want to get is inspired by the *Freedom Beyond Limits* message behind our organisation, and the words of our Freedom waiata. I felt a strong personal and emotional connection to those words the first time I heard them, and hope these words and meanings give strength and empower many more people.

"E rere, ki hea, puta atu, rere tonu.

Iti rearea, kahikatea, ka taea, ka taea.

True freedom is within, to live beyond our limits, and rise above challenges, with perseverance and commitment.

Like the tiny rearea bird, whose size is no limit, to it standing atop the tall kahikatea, we soar on the wings of freedom."

Save the date

6th Sept.
Freedom Campaign Launch, Stardome Observatory, Auckland

7th Sept.
Global DMD Awareness Day – online balloon promotion via Facebook

9th Sept.
Neuromuscular Child Health Seminar, Auckland

15th Sept.
FSHD Scientific Seminar, Auckland

16th Sept.
Cocktail Event & Charity Auction, Auckland

23rd Sept.
CMT Awareness Month, Seminar, Christchurch

29th Sept.
Nationwide Freedom Street Appeal

30th Sept.
LGMD Awareness Day – online promotion

Find out about these and other events in the regions at www.mda.org.nz

BRANCH news

Catching up with news from around the country

Canterbury

Our mid-winter lunch held at the Aspire Center was an amazing afternoon with over 40 members attending. Many thanks to Bonny and the rest of the team for putting this together. We are also grateful for the kind donations from our local community.

The Freedom campaign is fast approaching, and we need volunteers to help us collect donations at Bunnings, The Warehouse Riccarton and The Palms shopping mall on 29th and 30th of September. If you are able to lend us a hand during this time please call the office on 0800 463 222

We're looking forward to our children's camp on October 2nd - 5th of October for kids aged 8 - 18 years. We still have room for more and would love to take you with us to Mt Hutt. We have exciting activities planned including jet boating, camp fires and hot pools. Let us know if you want to join the adventure.

Remember to pop over to our Facebook page to keep up to date with events and photos.

mdacanty@xtra.co.nz

Enjoying the mid-winter lunch.

Northern

The annual MDN Family Camp in Ngaruawahia was a great success and was our biggest ever. There were plenty of activities to keep everyone amused including bumper boats, kayaks, a heated pool, craft activities, and model boats. All the mums received beautiful Mothers' Day gifts and we thank all of the contributors who made these goody bags possible.

Our coffee group in Hamilton continues to meet on the second Wednesday of the month at 10.30am, Café Impresso, Rototuna Shopping Centre. We thank Serena Leong-Teo for her amazing effort and commitment to running our North Shore coffee group for the past 5 years. The branch is now looking for a new coordinator and members keen to revitalize the group. Please let us know if you are interested.

support@mdn.org.nz

There was plenty to keep everyone busy at camp - and special treats for the mums.

Wellington

National Volunteer Week was the perfect opportunity to thank all the people who work so hard for the branch. Liz Mills (pictured with Office Manager Elizabeth McCallum) has been a volunteer for several years and was presented with a gift voucher donated by Caffiend, a café close to the branch office, and a box of chocolates, as a thank you.

Planning for the September Freedom Campaign is well underway. Our branch will be collecting in the following areas: Palmerston North, Hastings, Gisborne, Napier, Kapiti Coast (28th September), Wellington Railway Station, Upper Hutt. Lower Hutt and Porirua.

Keep an eye on our Facebook page more information about the following events:

- Funny Money Casino Evening in Hastings on 23rd September
- Movie Night at The Light House Cinema, Petone on 24th September
- Art Auction at Southwards Car Museum, Otaihanga, Kapiti Coast on 28th October N

office.mdawgtn@xtra.co.nz

Image credit: WellingtonNZ.com

Elizabeth McCallum and Liz Mills during National Volunteer Week; we're booked into the Lighthouse Cinema for a movie night.

Southern

The first face to face meeting of our new Southern Regions Branch Committee was held in Gore on Saturday 10th June. It was great to meet new members of the committee and to work on the governance priorities for our group.

The first Seasons for Growth seminar was held in Dunedin at the end of June, with 14 people taking part. The seminar was promoted through MDANZ networks, Facebook, emails and letters to members, newsletters and brochure drops.

It was a great success and lots of fun. All participants engaged with each other and after a slightly challenging start with technology issues, everybody participated and left with a greater understanding of the processes of grief and loss. Feedback from participants will inform our next seminars.

We are planning for a Dunedin based therapeutic group to start in October, and more introductory seminars in Invercargill and North Otago later in the year. To find out more, please get in touch with Fieldworker Jo Smith on 027 509 8775 or email joanne@mda.org.nz

We are looking forward to the Month of Freedom and have some exciting events planned including a pop-up café on Saturday 30th September in Dunedin Central. Any volunteers or offer of help for the Freedom campaign would be appreciated. N

joanne@mda.org.nz

Southern Branch Fieldworker Jo Smith with Branch Chairperson Robbie Verhoef.

We are family: Jessica, Gina, John and Heather.

Our family, our condition

Three generations of one family share their journey with CMT

"CMT is what makes me, me."

Heather Anderson, 48

I was diagnosed with Charcot Marie Tooth disease (CMT) about 10 years ago, by a specialist at Christchurch hospital. I was the first of now eight in my family, across three generations, to be diagnosed. At the time I remember asking my GP about CMT, she told me she really didn't know anything about the condition and suggested I look it up on the internet. Eventually I was sent to the pain clinic and they helped educate my doctor a bit, as well as sorting out medication to help with managing pain.

We guessed my dad had it, but at the time he didn't want to be tested. We were pretty sure my teenage daughter had it too.

I think it was about five years ago I went to Auckland for a holiday. While in Auckland I visited a friend and she said I'm sure your condition is covered under MDA and she gave me an *In Touch* magazine. During this time I was at a very low point in my life. Up till then I had felt like I was on my own, especially as my doctor didn't understand the condition. I couldn't wait to get home to Christchurch and make contact with the MDA Canterbury branch. After

a visit from the local fieldworker my daughter and I felt supported. We joined the local coffee group, and still enjoy this and other social gatherings held by the branch, as well as *In Touch* magazine and all of the information and support on offer.

Since then my sister, a niece, three cousins, my dad and my daughter have all been diagnosed with CMT. We are guessing dad's brother and their father also had CMT, we just didn't know about it then.

One of the challenges I have faced is losing a job that I loved, as my condition worsened. I worked at a rest home as an activities assistant. As the need came for me to use a walking stick and my workplace management said I couldn't drive residents because of the numbness in my feet, my job was on the line and I was sent packing. I loved working alongside the residents who were a big part of my life, so I still visit the rest home weekly as a volunteer.

When I was a child, I remember the way my uncle walked and as awful as it is, we would laugh and copy the way he walked. I now walk like that. I have a drunk walk where at times a leg will be in mid-step and it doesn't seem to know where it wants to go. I have had people say "If I didn't know better I would think you were drunk." My uncle used to be called 'bumble foot'; he would trip over nothing. Well, that's me and my dad now.

CMT also affects my hands, I drop things often. Dropping a pen is fine, but if it's a chopping board or pot of boiled potatoes, something is going to hurt! The achiness in my hands, lower arms, feet and lower legs is unbearable at times. My toes are curling up and my big toes hurt often, as shoes aren't made for hammer toes. People find it hard to understand the tiredness, not being able to stand for long, and the achiness when walking, because it isn't always obvious or visible. This is especially so for my daughter who uses no aides for walking.

I have an adult tricycle (thanks to MDA for their support in helping me get funding for this). I love my trike as it gives me independence and freedom. It enables me to get to my craft groups, to the library, I can do my own shopping and you would be surprised what I manage to transport in it.

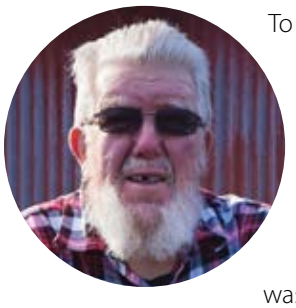
If I'm out on the bus, you may see me using a walker. It has a big crocheted mandala attached to the front that I made. I have had people come up to me and say, "I'm sorry you have to use that." It doesn't worry me anymore what people think. I feel my trike, walker, walking stick, my splints, kitchen trolley etc are aides to keep me living, they keep me on my feet (well most of the time) and I love them. They're not just for old people, they're aides for living and I tell people that.

Her bike means freedom for Heather.

I have an adult tricycle (thanks to MDA for their support in helping me get funding for this). I love my trike as it gives me independence and freedom.

My dad is 76 years old; he really struggles with CMT especially with not able to do all he used to. He's proud and doesn't like to ask for help, but he has started asking. I know he suffers with pain, which often comes out in cross words - he's a grumpy old man at times, but I love him just the same. He uses a walking stick and wears splints, because of drop foot and tripping. It has only been in his later years that CMT has started to affect him.

John Bolton, 76



To get a diagnosis, the doctor said, "Are you walking differently John?" Gina (my wife) said, "I can tell you what he has got, he has CMT". The doctor tried to test for any reflexes and I told him, "You're wasting your time I don't have any." The doctor did the blood test and it was sent to a geneticist.

It has been in the last five years CMT has started to show. It is the simple things like taking the milk from the fridge and lifting it onto the bench which has become difficult. I can no longer lift a hammer or use the ladder to do home maintenance. In the garden, it is easier to crawl around on my hands and knees than to get up and down all the time. I have a hard job doing up my buttons on my shirts – especially on a cold morning. It is not very often that someone tells me that my hands are warm. I have terrible balance. It is the fine motor skills, like using a pen to sign my name, which has become difficult.

I used to be the Santa at the Hub Hornby, however I can no longer do this, because I have trouble dressing myself.

Jessica Anderson, 24

I was diagnosed with CMT about six years ago. However, we guessed that I had it when I was in high school. It became impossible to get school shoes that fitted my feet. I mainly live in my shoes from New Balance as I can get 4E width (men's) shoes in a size 7 ½. I also have a high arch that collapses so I get sharp pains in my feet when I walk (especially when I don't have my prescription orthotics in my shoes). We tried getting specially made shoes, but I refused to wear them as they looked like clown shoes and made me look different, which was embarrassing for me as a young person.

Getting a diagnosis has been huge for me. I now have a physical reason for the symptoms I experience and it is good for me and others to know that I am not imagining them. I was the second one in my family to be diagnosed. It now made sense as to why from late primary school, I was tripping over my feet. People kept saying it was because I needed to grow into my feet as my feet were too big for my body, however at the time we didn't realise there was a physical reason for the problem. I had a period of breaking bones from tripping over my feet. It got to the stage where the ED nurses knew us by name and one joked that they didn't do Fly Buys – because we were like frequent flyers!

I am a third generation CMTer with a diagnosis and it seems to be getting worse with each generation because

Freedom for Jessica is getting out and about.

I am a third generation CMTer with a diagnosis and it seems to be getting worse with each generation because my symptoms started to show before my grandad's did.

my symptoms started to show before my grandad's did.

My feet are super-sensitive; the non-slip surface on the shower or around swimming pools hurts my feet. Whereas my mum can walk over anything and not feel it. That numbness may yet be in my future – I will have to wait and see. My feet ache as if I have been on them all day, and I have achiness when walking, I have been told off for not standing up to give up my seat to someone who looks like they need it more, or for using a lift and not walking up the stairs. This is hard because people make assumptions. My disability is hidden, I'm young and people just do not understand my condition.

CMT especially affects my hands and wrists. I cannot write for long without getting sore wrists. I'm left handed so maybe it doesn't help with the way I write (keeping my hand from smudging the paper). I often need to use wrist supports. My wrists often feel like I have sprained them without doing anything. People often ask what I have done to my wrist, to which I usually say, "Nothing, it's just sore." They don't get it.

I have poor circulation. During the winter I live in my fingerless gloves as my hands always feel cold. I have trouble keeping my feet warm, I have been known to wear two pair of merino socks and still have frozen cold feet, and it's even worse with my mum.

My balance is shocking. I can be walking along and all of a sudden my foot doesn't know where to go. It is a wonder I haven't been stopped before for looking drunk. My balance is worse when I am tired.

I remember laughing about grandad and how he walks when I was younger. We would say, "Here comes grandad." You could hear him slapping his feet. I now do the same, however this is worse when I am tired.

Freedom for me is riding my bike, getting me to where I need to go and saving money on bus fares, especially as I'm a poor student. However, I have to be very careful how I get on my bike as I have lost my balance when trying to get on. The cycling is great for my legs; however I find the jarring affects my hands. My hands quite often go numb and get pins and needles. I bike along flicking my fingers open and closed trying to regain feeling. Other people biking along must think I look bonkers.

But even with all of these challenges, I wouldn't change anything as CMT is what makes me, me.

Understanding the effects of Charcot-Marie-Tooth disease

The first New Zealand study into living with CMT

The Impact CMT study aimed to find out how many people are living with CMT, in New Zealand. Previously this information was only available from studies conducted overseas. The work was funded by Neuromuscular Research New Zealand to help us to understand the impact of this condition here in New Zealand.

What we did

This study was conducted in partnership with the Muscular Dystrophy Association, the New Zealand Neuromuscular Disease Registry and Auckland District Health Board. A team of researchers and clinicians got together to try and find every living person in the Auckland DHB region who had been diagnosed with CMT. Auckland was chosen because it has both neurogenetic and neuropathy clinics and because the DHB covers 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.

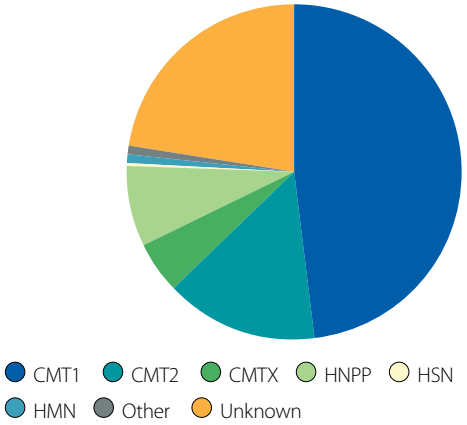
People living with CMT were identified through the Muscular Dystrophy Association member database, the NZ Neuromuscular Disease Registry, hospital records, neurologists, orthotics centres, the Genetic Health Service and Health Labs. We also received referrals from clinicians working in the community such as GP's, occupational therapists, podiatrists and physiotherapists, patient support organisations as well as advertising in local papers and on social media.

When a person was identified, we recorded information about their age, gender, ethnicity and diagnosis/genetic test results from their medical records. In addition to counting these numbers of people, we offered everyone we could contact a chance to complete an interview about how the condition affected them their lives.

What we found

We identified 237 people with a diagnosis of CMT living in Auckland. From this we can estimate that there are about 710 people living with CMT in New Zealand. There was an even gender split with (50.2%) being male. As expected, most people had subtype CMT1 (where the myelin surrounding the nerve is affected) and of these, most people (83.3%) had type CMT1A, which is caused by a duplication of the PMP22 gene. Just over half of the group had a genetic test, with two thirds of those tests providing molecular confirmation of diagnosis.

Numbers of people with different clinical types

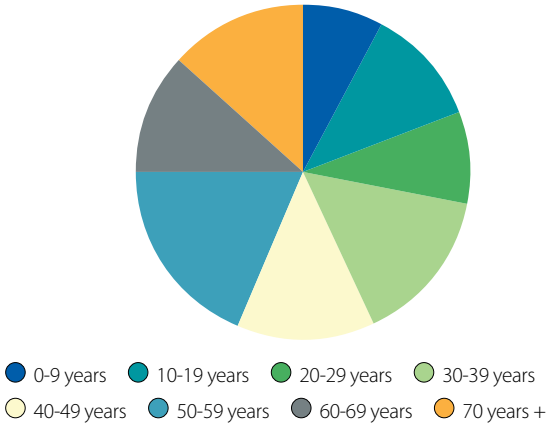


HNPP = Hereditary Neuropathy with liability to pressure palsies, HSN = Hereditary Sensory Neuropathy, HMN = Hereditary Motor Neuropathy

We identified 29 children (under 16 years of age) and 208 adults living with CMT. The proportion (12.2%) of

children identified is lower than overseas estimates. We found a remarkably constant prevalence at all ages, reflecting that CMT seldom foreshortens life.

Age distribution of people living with CMT



Symptoms

The most common symptoms for adults and children diagnosed with CMT were loss of sensation and weakness in legs and feet and/or hands and arms. Just over a third of people were experiencing moderate to severe levels of pain. Over half of the sample described experiencing trips or falls.

People's experiences

We asked people taking part in the study to talk about what the condition was like for them. People's experiences varied a lot with some people reporting minor impacts on their life, whilst others found managing everyday tasks challenging. Some key elements that were common throughout people's experiences as outlined below.

It's the small things

Many talked about the frustration of completing daily tasks and trying to reduce the impact on others. People described frequently missing out on activities as they did not want to be a burden on others.

"It's the day to day things that others take for granted that you think about… like going to open homes and not knowing how difficult they will be to get around or going on boats. I don't like to slow others down so sometimes I choose to sit out of an activity if I think my CMT might be an issue."

Invisibility of symptoms

People described that as not all symptoms were visible to others, this often made it more difficult to deal with and hard for others to understand.

"On the surface you look quite normal so people expect you to do everything and don't understand the fatigue which is part of the condition."

People were also concerned about what others thought of them because of this;

"As most people wouldn't know I have it [CMT] by looking at me, I find it difficult as I think others may perceive me as lazy. Like not being able to join others for walks or hikes or sports. I find that difficult. Also I find keeping up with people when walking down the road hard, I tend to avoid walking with people who aren't aware of my condition."

"As most people wouldn't know I have it [CMT] by looking at me, I find it difficult as I think others may perceive me as lazy."

Need for physical supports

People talked about the fact that it was hard to access physical supports in a timely manner as their condition changed. Many felt the design of physical supports could also be improved. For example:

"The foot braces are ugly and their design hasn't changed in 10 years."

The visible nature of physical supports also had an impact on self-esteem.

"CMT has affected my self-esteem because I see myself with a walking stick and it feels unattractive."

More awareness needed about the condition

The most common thing people referred to was the lack of information and knowledge about the condition. Friends and family, as well as community health professionals also noticed a lack of information.

Image credit: facebook.com/CMTAssociation

"I have a chronic condition - there's a level of anxiety about what the future holds ... What's down the road?"

"Feels like health professionals who are not specialised in the field do not know anything about the condition. There needs to be more awareness and education for primary health care professionals on CMT."

The psychological impact of living with CMT

People's experiences of the progression of their condition varied a lot, with some experiencing little impact on their lives whereas others found the gradual loss of abilities difficult, and the uncertainty about the future difficult to deal with.

"I think there's a huge psychological impact. I don't know that anyone's offered support in that regard. I think that's huge. I have a chronic condition - there's a level of anxiety about what the future holds. I don't think this questionnaire touches on that much (the psychological strain and stress). What's down the road?"

Experience of health care services

Many described initial challenges in getting a diagnosis or accessing services. Once connected with health care services, however, many reported a positive experience and feeling well supported. It was highlighted that follow up outpatient visits were needed as the condition changed. For example, a parent of a child with CMT said:

"When he was first referred it went nicely, but now he would like to have some follow-up, for example, a clinic run which sets up appointments every 4 to 5 years. Or a CMT-dedicated repository to answer questions. Initial diagnosis is fantastic but follow-up is weak"

What happens next

There is a wealth of information from this study, so we will be continuing to analyse the data and will present more findings as they become available. We hope that the findings of this research will improve our understanding of CMT and increase the support available to adults and children living with this condition.

We would like thank everyone who contributed to this study - we couldn't have done it without you!

Alice Theadom

What does a TAiQ powerchair

have in common with..

TA the new standard

see the answers below and ask us for a hot lap

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Answers: TA versus other powerchairs

As low as a Ferrari. All TA powerchairs have the lowest ground to seat height at only 38cm/15" with electric hilow seat function as standard. Anytime, anywhere sit with ease under a table or desk.

A ride as soft and sweet as a marshmallow. Experience the difference for yourself over uneven terrain or kerbs.

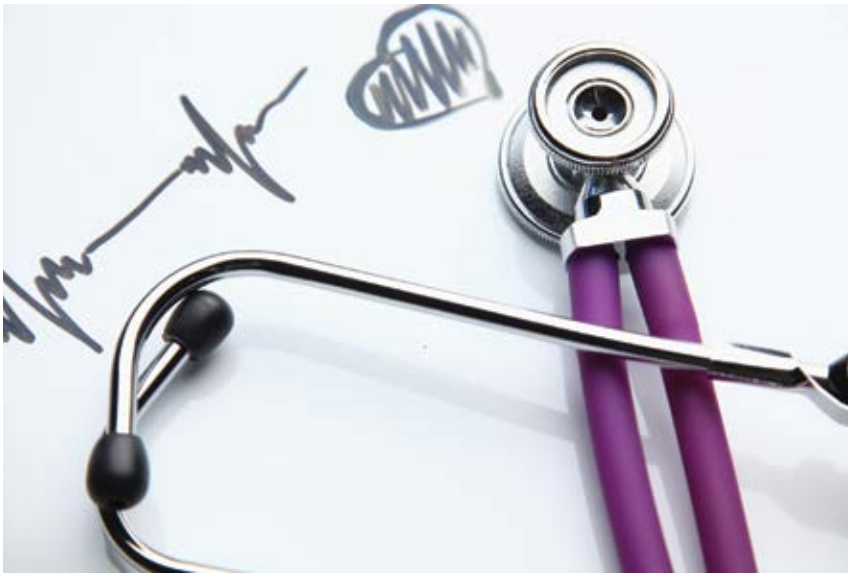
View our videos at www.mortonperry.co.nz/mobility-ta-iq.html or scan the QR code with your smartphone.

Update on Raxone for DMD

Slowing respiratory decline in DMD.

Raxone is a drug developed by Santhera Pharmaceuticals which aims to slow down respiratory decline in Duchenne muscular dystrophy (DMD). The active substance in Raxone is called idebenone, an anti-oxidant that improves energy production in mitochondria.

Santhera recently announced that The European Medicines Agency (EMA) Committee for Medicinal Products for Human Use (CHMP) has granted approval of Raxone via the Early Access to Medicines Scheme (EAMS) to treat the decline in respiratory function in people with DMD from the age of 10 years who are currently not on glucocorticoids. The company is still investigating whether Raxone could also benefit those taking steroids in a study called SIDEROS. Ⓡ



Blood biomarkers for myotonic dystrophy (DM)

Maintaining heart health.

Cardiac troponin (cTnI) is a heart protein that leaks into the blood if the heart becomes damaged. The cTnI blood test is often carried out in hospital emergency departments for people with chest pain, as a very high level can suggest that someone is having a heart attack.

Dr Mark Hamilton and colleagues in the UK recently investigated whether much smaller changes in cTnI levels can give useful information about heart health in a group of 117 people with myotonic dystrophy (DM) attending routine outpatient clinics. Results were variable with nine individuals having excessively high cTnI values.

Although small, the study suggested that blood biomarker tests, such as the cTnI test, maybe a useful addition to annual ECG screening for people with DM. They could help to identify those who should have more detailed heart checks such as echocardiograms or closer follow-up.

The researchers hope that this will lead to larger studies exploring heart biomarkers in DM, working towards a more harmonised approach to cardiac care. Ⓡ

Resolaris in FSHD

Improving muscle strength.

Resolaris is a protein developed by aTyr Pharma that aims to reduce inflammation by altering the body's immune response. aTyr Pharma has been conducting Phase I and II trials in young adults aged between 16 and 20 years of age with early-onset facioscapulohumeral muscular dystrophy (FSHD), diagnosed before 10 years of age, to test safety and efficacy of Resolaris over 12 weeks, results of which have been recently announced.

The results showed that Resolaris was safe and well-tolerated, including at the highest dose. 63% of participants showed an improvement in muscle strength and 67% reported an improvement in their quality of life.

Resolaris now has Orphan Drug Designation in the USA for the treatment of both FSHD and limb girdle muscular dystrophy (LGMD) which will help with the development and marketing of this drug.

Preliminary results of SUNFISH trial in SMA

RG7916 is an oral drug developed by PTC Therapeutics that could be a potential treatment for spinal muscular atrophy (SMA). It aims to alter splicing of the SMN2 gene, in order to make it more active and produce more SMN protein. It is currently being tested in children and young adults with SMA type 2 and type 3 in a phase 2 study called SUNFISH.

PTC Therapeutics recently presented preliminary results at the Cure SMA conference, which show that RG7916 increased activity of the SMN2 gene in a dose-dependent manner. No adverse events have been reported so far, suggesting that the drug is safe and well-tolerated.

The SUNFISH study is ongoing and consists of two parts. Part 1 is evaluating safety, pharmacokinetics (blood levels across time) and pharmacodynamics (what the drug does in the body). It will also determine the dose of that will be used in Part 2. Part 2 aims to assess the efficacy of the drug by measuring participants' motor function.

RG7916 is also being tested in phase 2 clinical trials called FIREFISH (type 1 patients only) and JEWELFISH (patients with type 2 or 3 that took part in previous trials). It is being developed as part of a joint research programme between PTC Therapeutics, Roche and the SMA Foundation.

Drug for congenital myotonic dystrophy

Drug receives Fast Track designation.

AMO Pharma is a pharmaceutical company that has developed a drug called tideglusib or AMO-02 for the treatment of congenital myotonic dystrophy. AMO-02 inhibits an enzyme called glycogen synthase kinase 3ß (GSK3ß), which is over-active in people with congenital myotonic dystrophy. Preclinical research has shown that inhibiting GSK3ß can increase muscle strength and decrease myotonia in mouse models of the condition. AMO Pharma is currently testing the safety and effectiveness of AMO-02 in a phase 2 trial.

The US Food and Drug Administration (FDA) has granted Fast Track designation to AMO-02 which will speed up its development and enable faster access to it. Fast Track designation is a special status that helps to accelerate the development and review of drugs for serious health conditions. Drugs that receive Fast Track designation are eligible for more frequent meetings and written communications with the FDA, as well as accelerated review and priority approval processes. R

Weak feet and walking, it's in the shoes

The best footwear for kids with CMT.

Rachel Kennedy, a physiotherapist and PhD candidate at the Melbourne School of Health Sciences, conducted a study to see if there was a difference in walking speed, cadence, step length and step width in children and adolescents with CMT when walking in optimal footwear, compared to walking in suboptimal footwear (such as jandals or Crocs).

In optimal footwear children walked faster by 7 cm/second ($p<0.002$).

Footwear type has a moderate effect on gait speed ($d=0.41$).

Step length was 3 cm longer in optimal footwear ($p<0.002$).

Children and adolescents with CMT walk faster with a longer step length

when wearing well-fitting "optimal" footwear compared to "suboptimal" footwear.

In a condition where problems with walking and balance have a major impact on quality of life, footwear type and fit matters. R

New biotech launched to advance LGMD gene therapies

A new biotechnology company called Myonexus Therapeutics is developing gene therapies for limb girdle muscular dystrophy (LGMD). Myonexus' gene therapies use an adeno-associated virus (AAV) to deliver the desired gene into the body. Myonexus' pipeline includes three clinical-stage programmes for LGMD 2D (alpha-sarcoglycanopathy) and LGMD 2B (dysferlinopathy) which are currently being tested in phase 1 trials in the USA. The third trial testing the LGMD 2E (beta-sarcoglycanopathy) gene therapy is expected to begin in November 2017. The company will also advance another two preclinical programmes for LGMD 2C (gamma-sarcoglycanopathy) and LGMD 2L (anoctaminopathy). R

Is resistance exercise ok for kids with CMT?

Slowing down muscle weakness.

A randomised, double-blind, sham-controlled trial was carried out across the Sydney Children's Hospitals Network (NSW, Australia) to assess the safety and efficacy of progressive resistance exercise for foot dorsiflexion weakness in children with CMT disease.

Sixty children aged 6–17 years with CMT disease with foot dorsiflexion weakness were randomly assigned to receive six months of progressive resistance training or sham training (negligible non-progressed intensity), using an adjustable exercise cuff to exercise the dorsiflexors of each foot. Training was carried out three times per week on non-consecutive days with 72 sessions in total.

The primary efficacy outcome was the difference in dorsiflexion strength assessed by hand-held dynamometry from baseline to 6, 12, and 24 months. The primary safety outcome was the difference in muscle and intramuscular fat volume of the anterior compartment of the lower leg assessed by MRI.

Results showed that six months of targeted progressive resistance exercise slowed down long-term progression of dorsiflexion weakness without detrimental effects on muscle morphology or other signs of overwork weakness in children with CMT. (R)

Myostatin inhibition for SMA

Potential treatment supported.

Biotechnology company Scholar Rock recently announced that its myostatin inhibitor, SRK-015, will be developed as a potential treatment for spinal muscular atrophy (SMA).

Myostatin is a protein that limits muscle growth and stops our muscles from becoming too big. Blocking myostatin allows the muscles grow and become stronger, which could potentially be beneficial for people with muscle-wasting conditions such as SMA.

Scholar Rock presented new preclinical data for SRK-015 at the Cure SMA conference. This showed that SRK-015 selectively blocks myostatin and does not interact with other closely related proteins. This selectivity is important as it may reduce the risk of side effects of the drug.

The company tested SRK-015 in non-human primates and found that it increased their lean body mass. It was also tested in a mouse model with SMA, in combination with a SMN therapy that targets the underlying genetic cause of SMA. This significantly improved the muscle strength of the mice and was more effective than the SMN therapy alone.

Scholar Rock is aiming to commence clinical testing of SRK-015 in mid-2018. It intends to develop SRK-015 both in combination with SMN-based therapies and as a single therapy for people with certain types of SMA. (R)

Charcot-Marie-Tooth disease

Understanding our most common condition.

What is Charcot-Marie-Tooth disease?

Charcot-Marie-Tooth disease (CMT), also known as hereditary motor and sensory neuropathy (HMSN), comprises a family of genetic conditions that mainly affect the motor and sensory nerves, which run from the spinal cord down the arms and legs.

CMT is named after the three physicians who first described it in 1886, Jean-Marie Charcot, Pierre Marie, and Howard Henry Tooth. CMT is a relatively common rare disorder affecting both males and females from childhood or adulthood depending on the type. It's estimated that CMT affects 1 in approximately 2,500 people.

The loss of nerve function is often accompanied by tingling and burning sensations in the hands and feet.

What causes CMT?

CMT is caused by alterations in genes that produce proteins involved in the structure and function of either the peripheral nerve cell (axon), or the myelin sheath that wraps around the axon to insulate it and in normal circumstances allows the signals to travel faster and better.

What are the different types of Charcot-Marie-Tooth Disease?

There are many forms of CMT. Due to the increase in knowledge of the various genetic causes of CMT, the way the different types are described and classified is being re-thought. The most common form is CMT is called CMT1A and is caused by a duplication of a gene called PMP22. It's inherited in an autosomal dominant way, meaning that a child with CMT1A will have one parent with the condition. CMT1A affects the myelin sheath surrounding the nerve. There are other forms of CMT1 and what they all have in common is that they affect the structure and function of the myelin sheath.

Symptoms of CMT1A usually become apparent during childhood. Children with this condition are often slow runners, develop high arches, hammer toes and often require orthotics (braces) for ankle support as the ankles weaken and become prone to sprains. Later, often ten years or more after the onset of foot and leg problems, varying degrees of hand weakness can occur resulting in problems holding pens, grasping, performing fine motor tasks such as doing up buttons or zippers. Problems with balance because of ankle weakness and loss

of proprioception are common, although most people remain mobile throughout life, and life expectancy is normal.

CMT2 is caused by abnormal genes involved in the structure and function of **axons** and is divided into subtypes on the basis of which gene is involved. The clinical picture is similar to CMT1 with distal weakness, muscle atrophy, sensory loss and foot deformities, but is often more variable between and even within families. Some types of CMT2 also cause deafness or eye problems.

Other features of CMT include:

- The loss of nerve function is often accompanied by tingling and burning sensations in the hands and feet. This usually causes little more than mild discomfort, but some people experience severe neuropathic pain and require medication to control it.
- Loss of nerve function in the extremities can also result in sensory loss. The sense of touch is diminished, as is the ability to perceive changes in temperature, and patients may unknowingly injure themselves. They can be unaware of having developed ulcers of the feet or of cuts or burns on the hands. Sensory loss in CMT patients may also be associated with dry skin and hair loss in the affected areas.
- Many patients are extremely sensitive to the cold or even to temperatures a few degrees lower than normal. CMT results in the loss of insulating muscle mass, which, combined with reduced muscular activity and circulation, can leave patients with chronically cold hands and feet. Impairment of the normal circulatory process can also result in swelling (oedema) of the feet and ankles.
- Deep-tendon reflexes, such as the knee jerk reaction, are lost in many patients, and is of diagnostic importance. Some people with CMT also have tremor (usually of the hands) and the combination of tremor and CMT is sometimes referred to as Roussy-Levy Syndrome.
- Scoliosis or mild curvature of the spine can also occur, often in puberty and tends to be most common in people with early onset of gait abnormalities. Hip dysplasia also affects a small number of CMT patients at an early age.

Deep-tendon reflexes, such as the knee jerk reaction, are lost in many patients, and is of diagnostic importance.

Diagnosis of Charcot-Marie-Tooth disease

Diagnosis of CMT begins with a standard patient history, family history, and neurological examination. People will be asked about the nature and duration of their symptoms and whether other family members have the disease. During the neurological examination a physician will look for characteristic features of the disease, and will then order diagnostic testing, which may include the following.

Electrodiagnostic Testing

Electrodiagnostic testing usually includes a nerve conduction velocity test (NCV), which measures the strength and speed of electrical signals moving down the peripheral nerves. Delayed responses are a sign of demyelination, and small responses are a sign of axonal involvement – some types of CMT show mixed both axonal and demyelinating signs. An electromyography (EMG) is another type of electrodiagnostic test, which measures the electrical signal's strength in the muscles of the arms or legs.

DNA Studies

These molecular genetic tests, usually performed on white blood cells extracted from a blood sample are available to test for some of the common genetic causes of CMT, including PMP22 duplications and deletions, MPZ and GJB1. A positive genetic test can provide definitive diagnosis and provide useful information for family planning, hence genetic counselling is highly recommended. However, a negative result does not rule out CMT as many forms are not yet able to be tested by single-gene DNA testing. Neuromuscular gene panels are increasingly being used once the common causes have been excluded.

CMT is managed symptomatically via a number of therapies depending on the needs of the individual.

Management of Charcot-Marie-Tooth disease

CMT is managed symptomatically via a number of therapies depending on the needs of the individual.

These include:

- **Podiatry:** for the care and monitoring of foot problems.
- **Orthotics** for the manufacture and fitting of braces and customised footwear.
- **Orthopaedic surgery** to traighten toes, lengthen heel cords or lower arches;
- **Physiotherapy and occupational therapy** to design exercise programmes to strengthen muscles or learn about energy conservation, as well as provide assistive devices and equipment to be able to continue day to day activities as independently as possible.
- **Dietitian visits or dietary advice** to maintain a healthy, balanced diet and weight.
- **Regular medicines reviews:** some medications should be avoided by people with CMT, as they are contra-indicated and can seriously worsen the condition or cause severe side-effects. The full list of medications to be avoided by people with CMT is available on the MDANZ website or can be obtained by contacting MDANZ. ®

Understanding orthotics

We asked Mark Leonida from the Orthotic Active Clinic to answer some commonly-asked questions.

Seeing an orthotist can make a big difference to children, young people and adults, presenting in both the early stages of CMT and at advanced stages of their journey.

Q. What is orthotics?

The purpose of orthotics may be to immobilise, support, prevent abnormality, correct abnormality or to assist or restore function. This is done through fitting various orthoses. Examples are: supports, corsets, braces, calipers, cervical collars and slings. Other examples are surgical footwear and footwear items (insoles, arch supports, shoe raises, heel cups).

Q: Do I need a referral to see an orthotist?

Your GP, specialist or other health professionals can refer you privately. You may also self refer. However, for complex problems, we recommend you first see a medical specialist. There is a pathway to access Ministry of Health Orthotic Services for you and your whānau.

Q: What can I expect at an appointment?

Once a referral has been received, the client will be assessed by a qualified orthotist, and a treatment plan and orthoses formulated to best treat the current client problems. The orthoses can be selected or custom made and applied, the functionality assessed and fine-tuned, for optimisation and to allow goals to be achieved.

Using state of the art casting, 3D scanning and CNC manufacturing techniques, our seven branches around the country are able to provide an orthotic solution for every individual situation and requirement.

The Orthotic Active clinic which is the private and ACC provider, also provides higher end orthoses such as functional electrical stimulation and tuning of gait patterns. With new technological breakthroughs and product lines, this has the potential to significantly enhance and improve quality of life.

Q: What changes will occur as my condition progresses?

Currently, I have several CMT clients on my case load and these range from younger teenagers with early diagnosis, to middle aged and more senior clients. The intervention will change with the condition and therefore constant review is required. A simple support early on in the condition may change to an orthoses that has to replace function later in life to maintain activity level, replacing muscle function, as well as maintaining alignment.

For example, let's start a journey with an early referral for a teenager with CMT. Younger people may only require a simple supportive insole to cater for the mild neurological presentations and cavus nature (high arch) due to the changes in the musculature and structure of the foot during growth. This may progress to an ankle foot device later to support weaker ankle structure and prevent repeated ankle sprains. Later, as the condition progresses, the client may not be able to lift their foot when they swing their foot through when walking. And later still, require an ankle foot orthoses (AFO) that gives energy return properties, such as a carbon fibre device that will give them spring and push-off whilst their weight is on the floor, and lift their foot up preventing foot drop when swinging their leg through.

Should the client require custom footwear due to the changes in their foot shape and function, a full custom footwear solution is available, using the latest scanning techniques, achieving a high quality modern design shoe, which is both functional and aesthetically pleasing to the user.

The Orthotist's involvement is to be on the journey along with their client.

www.orthotics.co.nz

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Orthotic Centre Active

GUEST
speakers

On holiday with a friend in Melbourne.

Taking time to relax

OLIVIA SHIVAS

Life can get busy and as much as I love having a full life, I think it's so important to take a break when things get overwhelming.

I knew I had a hectic period coming up at work so I planned in advance to have a holiday afterwards. It was motivating to have something to look forward to. After a few months of late nights at work, near-impossible deadlines, and finally being satisfied with the projects I had completed, I turned off my work email notifications and headed to Melbourne.

... this place was full of fascinating accounts of living through war times; it really makes you appreciate the way we live now.

My friend Victoria and I stayed at a hotel right in the city. It was within walking distance to everything we wanted to do and see, and the hotel had great wheelchair access. These two things are really important to me when I'm travelling! We were only there for five days, but planned to see a few sights, go shopping and drink lots of coffee. One of the highlights for me was visiting St Patrick's Cathedral. This wasn't on our to-do list, but Victoria saw a tall steeple in the distance and we decided to follow it. What we came across was an incredible Gothic Revival-style church. I love stained glass windows and have another level of appreciation for them now. There's also a sense of peace I feel in an old church like that, which was just what I needed at the time.

Another highlight was visiting the Shrine of Remembrance, a memorial to Australians who have served in the war. From the outside, this building is stunning and has a wide flight of steps at the front - and in the back of my mind I'm always thinking, 'Is this place accessible?' I was able to get to all the main galleries and the Sanctuary, where they have a three-minute memorial service every day. Although there were only stairs to get to the balcony at the very top of the building, I still enjoyed exploring the areas I could get to. I love storytelling and hearing other people's stories, and this place was full of fascinating accounts of living through war times; it really makes you appreciate the way we live now.

On the last day of my holiday, I was just starting to think about work - it made realise how much I enjoy and appreciate my job. Coming back home, I felt refreshed and (surprisingly) ready to dig into some new projects at work. So as we head into a new season, think about what you enjoy doing in life and make time to actually do it!

Olivia is the Rangatahi representative on National Council. She lives with central core disease and has a passion for seeing young people reach their full potential. Olivia has a Bachelor of Communication Studies and works at Attitude Pictures, a TV production company that promotes the stories of people with disabilities.

Sports and disability

DR HUHANA HICKEY

Q: I'm hoping to participate in a major sporting event this summer. Considering that I use a wheelchair, what can I expect from the event organisers and how can I make sure my needs are met?

There is a big difference in support and resources/funding for those who have ACC related conditions and those with Ministry of Health funding. When I did wheelchair cycling, I had to buy my own chair to compete. Often there were no disability toilets available at the finish line, so I had to leave immediately. It meant I couldn't participate fully. When I played wheelchair rugby, I could use loaned chairs and that helped heaps.

The concept of full inclusion is the ideal, but the reality is unless you have resources, you need to find funding. There are some networks that have loan wheelchairs or adapted equipment to use, but if you want to get into sport seriously it's always ideal to get your own equipment. There are some avenues, such as lotteries, but they are very limited. There is funding from some of the disability organisations, such as the Jubilee Trust through CCS Disability Action. There is also the Halberg Trust, www.halberg.co.nz . They work primarily with young people with disabilities to encourage them into sports. Our nephew, who is seven and lives with spina bifida, had the fun of attending a fundraiser for them and cycled in a wheelchair cycle where he is showing some great talent. I have personally seen a lot of young people experience sports in an inclusive way through the Halberg trust. Their website is excellent. Contact them for ideas, advice and support.

... the only way we can be fully included is to challenge the rules set up to exclude us.

If you are an adult with high aspirations, consider registering your interest with Parafed or Paralympics New Zealand. This will provide you with networks and supports to achieve in your chosen sport.

If you want to attend a mainstream sports event, chances are it will not meet the needs of all the community, and will often fall short. In fact, some events exclude disabled participants because of concerns over health and safety. Sometimes we have to advocate for our inclusion. I would encourage people to do that, because the only way we can be fully included is to challenge the rules set up to exclude us. Paralympian Sophie Pascoe has argued for full inclusion to compete alongside non-disabled athletes. If an elite athlete is struggling to get recognition, then imagine what it's like for the average community member with access needs. However, if a sporting organisation is keen to engage and work out how to include you, then also get your local council sports advisors to assist. Groups such as Sport Waikato and Counties Manukau Sport can help you to achieve inclusion and find funding. Paralympics New Zealand www.paralympics.org.nz also has useful tips.

When someone says they cannot accommodate you, tell them it's their attitude limiting your potential, not your disability, and encourage them to reconsider.

Dr Huhana Hickey MNZM has a background in human rights and disability law, she is currently a post doctoral research fellow at AUT where she is studying the health and disability needs of whānau hauā. She remains committed to ensuring all persons with disabilities and their whānau know of and have access to their rights.

GUEST speakers

Members of the co-design group at work.

Future direction

RONELLE BAKER

Transforming the disability support system to enable better lives.

In March 2017, a 13-person co-design group was formed to look at the disability support system and identify ways it could be redesigned to offer more flexible and responsive service options that enable disabled people to have better lives. The group includes disabled people, family representatives, representatives from service agencies and government officials.

The process is being supported by the Associate Minister for Health and the Minister for Disability Issues. The Ministry of Health is the lead government agency, with the Ministry of Social Development and Ministry of Education also playing a role.

The aim for New Zealand's disability support system is that it will be re-designed to align with the vision and principles of the Enabling Good Lives (EGL) model. EGL has been trialled in the Waikato and Christchurch and is an approach that gives disabled people and their families more choice and control over the kinds of supports they receive. A review of the EGL trials/demonstrations has shown the approach can improve quality of life and outcomes for disabled people and their families.

In addition to the EGL model, the co-design group were also asked to consider a social investment approach in their redesign work. A social investment approach will bring evidence and results into the picture, and encourage early investment to improve long-term outcomes for disabled people.

The co-design group have participated in 10 intensive workshop style meetings, across a three month timeframe from April - June. Cabinet will soon consider their design recommendations for the disability support system, and once agreed, the new service model will be trialled in the Mid-Central region (around Palmerston North).

During this time, the Waikato and Christchurch EGL demonstrations will continue, and people involved in these trials will not have their services disrupted.

The future of the New Zealand disability support system will be influenced by the outcomes of this transformation process. To find out more, or get involved, you can visit the Enabling Good Lives or Office for Disability Issues websites.

These websites are;

www.odi.govt.nz/nz-disability-strategy/other-initiatives/transforming-the-disability-support-system/faqs-transforming-the-disability-support-system/

www.enablinggoodlives.co.nz/

You can also contribute directly to the co-design process by submitting ideas or suggestions to STfeedback@moh.govt.nz.

To help people become more familiar with the EGL model, a range of workshops are being offered by NZDSN. The next workshops are as follows:

30 August, Nelson

27 September, Christchurch

There is a fee to attend these workshops and MDANZ can help with this cost. Please contact us at info@mda.org.nz or 0800 800 337 if you would like support to attend.

Ronelle is the chief executive of the Muscular Dystrophy Association of New Zealand.

Freedom beyond limits

DR. RICHARD ROXBURGH

What does that phrase mean to a neurologist?

My immediate thought was that this was about compensating someone with a physical or cognitive disability, so they could take part in everyday human events in the same way as someone without disabilities do.

The most obvious example would be giving a wheelchair or adapted transport to someone who can't walk because of muscular dystrophy, so they can still be mobile, and access public places and the homes of their friends and family. Another example is a child with dyslexia having someone to be their reader/writer.

Yet, in a way we all have limits. I will never run like Usain Bolt or think like Stephen Hawking, so I think Freedom Beyond limits is about letting someone realise what capabilities and dreams they have. The child in the above example might have a huge imagination and capability in one cognitive domain, and this needs to be set free by compensating in another.

That means that our society needs to cater to the individual. I frequently write letters to funding agencies asking them to see beyond their prescribed guidelines to do what will really make a difference for someone. Unfortunately, I have to admit I don't often win. One example particularly grieves me. I know an independent young man who lives in a downstairs flat, with his parents living on the same property upstairs. The funders will only provide wheelchair access for the young man to his own house yet his life would be hugely enriched by being able to get wheelchair access to the family home, upstairs. The other frustrating scenario is the ongoing saga about carers not being funded to come into hospital with their clients.

Of course there is a danger that we can become constantly aware of our boundaries, and not rejoice in what we can do - easier for me to say than many. But I do often see that sometimes the fight to cope – not only through adapting our environment, but also through internal resilience – brings qualities the rest of us have no knowledge of. This can be taxing, even overwhelming, but I have also seen it be transformative. It is my privilege as a doctor to hear such stories on a frequent basis.

There are also other limits that I'm aware of. In the daily busyness that is hospital practice, it is difficult not to keep doing the same old thing, without believing that changes in delivery of care - and even cures - are possible. For me, the recent approval of a drug for the treatment of SMA gives me hope that neurogenetic diseases are indeed, the most curable, incurable diseases.

... I do often see that sometimes the fight to cope – not only through adapting our environment, but also through internal resilience – brings qualities the rest of us have no knowledge of.

We need to continue to challenge what our mind thinks is possible. Beyond being stuck with what we have. Beyond, "We're just a small country". Beyond "We don't have enough money".

Dr. Richard Roxburgh FRACP PhD is a Consultant Neurologist at Auckland Hospital.

Muscular Dystrophy New Zealand

About us

MDANZ is a trusted source of specialist information and provides a range of free services and practical support for individuals, families and whānau with lived experience of rare neuromuscular conditions.

The Muscular Dystrophy Association of New Zealand Inc., commonly known as MDANZ, began in the late 1950. Since then MDANZ has broadened its scope to support many other neuromuscular conditions. We are proud to have Judy Bailey and Dame Susan Devoy as our longstanding patrons.

Our unique governance structure ensures leadership of the organisation by individuals and family members with lived experience of a neuromuscular condition. We have four regional branches that are supported by the National Office based in Auckland.

We want New Zealanders with lived experience of neuromuscular conditions to experience freedom of choice in a responsive society.

To achieve this mission, we provide;

- Free information and advice, through our website, an 0800 info line and in paper booklet form
- A nationwide fieldworker service for personalised support
- Free loan of resources, such library books, recreational beach chairs and cough assist machines
- Funded support for counselling
- Discretionary funding for life enhancing resources not covered by government
- A high quality quarterly magazine to inform and inspire our membership and broader communities of support
- Funding for neuromuscular research and a mechanism to help New Zealanders to access clinical trials and new treatments
- Education workshops for members, health professionals, schools and others
- Advocacy and lobbying at a community or national level
- A platform for support groups and peer to peer networking

MDANZ is a registered charity and relies almost entirely on donations from the public, trusts and other businesses/ organisations to continue its work in the community.

Our Team

Ronelle Baker
Chief Executive

Miriam Rodrigues
Programme and Service Advisor

Brian Hadley
Accountant and Business Manager

Chris Light
Member Resource Assistant

Miriam Hanna
Information and Resource Coordinator

Amanda Lam
Accounts Assistant

Northern Branch


Fieldworkers: *Darian Smith and Kate Longmuir*
Office Manager: *Denise Ganley*
Ph: 09 415 5682 or 0800 636 787
Email: support@mdn.org.nz

Wellington Branch


Fieldworkers: *Dympna Mulroy and Penny Piper*
Office Manager: *Elizabeth McCallum*
Ph: 04 5896626 or 0800 886 626
Email: elizabeth@mda.org.nz

Canterbury Branch


Fieldworkers: *Paul Graham and Marty Price*
Office Manager: *Gemma Foulds*
Ph: 03 377 8010 or 0800 463 222
Email: mdacanty@xtra.co.nz

Southern Branch

Fieldworker: *Jo Smith*
Ph: 03 486 2066
Ph: 0800 800 337
Email: joanne@mda.org.nz

Council Representatives

If you want issues brought to National Council meetings, talk to your branch representative. They have the responsibility to raise your issues at National Council meetings and to make sure you are heard. Your branch representatives and their contact details are as follows:

Northern Branch

Trevor Jenkin. Ph: 021 267 4380
Email: trevor.jenkin@gmail.com

Wellington Branch

Annelize Steyn. Ph: 021 480 108
Email: kilmarnock.annelize@gmail.com

Southern Branch

Robbie Verhoef. Ph: 021 044 9437
Email: robbie.verhoef@yahoo.co.nz

Canterbury Branch

Warren Hall. Ph: 03 329 4390
Email: warrenjh@xtra.co.nz

Conditions covered by MDANZ

Muscular Dystrophies:

Becker Muscular Dystrophy

Congenital Muscular Dystrophies and Congenital Myopathies

Distal Muscular Dystrophy

Duchenne Muscular Dystrophy

Emery-Dreifuss Muscular Dystrophy

Facioscapulohumeral Muscular Dystrophy

Limb-Girdle Muscular Dystrophy

Manifesting carrier of Muscular Dystrophy

Myotonic Dystrophy

Oculopharyngeal Muscular Dystrophy

Diseases of the Motor Neurons:

Spinal Bulbar Muscular Atrophy (Kennedy's Disease and X-Linked SBMA)

Spinal Muscular Atrophy - all types including Type 1 Infantile Progressive Spinal Muscular Atrophy (also known as Werdnig Hoffman Disease)

Type 2 Intermediate Spinal Muscular Atrophy

Type 3 Juvenile Spinal Muscular Atrophy (Kugelberg Welander Disease)

Type 4 Adult Spinal Muscular Atrophy

Hereditary Spastic Paraplegias (HSP)

- all types:

Also called Familial Spastic Paraparesis

Leucodystrophies

- all types.

Metabolic Diseases of muscle *- all types including:*

Acid Maltase Deficiency (also known as Pompe's Disease)

Debrancher Enzyme Deficiency (also known as Cori's or Forbes' Disease)

Mitochondrial Myopathy (including MELAS, MERRF, NARP and MIDD)

Phosphofructokinase Deficiency (also known as Tarui's Disease)

Phosphorylase Deficiency (also known as McArdle's Disease)

Diseases of the Peripheral Nerve:

Charcot-Marie-Tooth Disease (CMT) (Hereditary Motor and Sensory Neuropathy) - all types

Dejerine-Sottas Disease (CMT Type 3)

Hereditary Sensory Neuropathy

Inflammatory Myopathies:

Dermatomyositis

Inclusion Body Myositis

Polymyositis

Diseases of the Neuromuscular Junction:

Congenital Myasthenic Syndrome

Lambert-Eaton Syndrome

Myasthenia Gravis

Myopathies *- all types:*

Andersen-Tawil syndrome

Central Core Disease

GNE Myopathy

Hyperthyroid Myopathy

Hypothyroid Myopathy

Myofibrillar myopathy

Myotonia Congenita (Two forms: Thomsen's and Becker's Disease)

Myotubular Myopathy

Nemaline Myopathy

Paramyotonia Congenita

Periodic Paralysis

Inherited Ataxias:

CANVAS

Friedreich Ataxia (FA)

Spinocerebellar Ataxia (SCA)

Neurocutaneous Syndromes *- conditions affecting the brain and the skin:*

Central Cavernous Hemangioma

Neurofibromatosis Type 1

Neurofibromatosis Type 2

Schwannamatosis

Tuberous Sclerosis

Von Hippel Lindau Syndrome

Should you have a query regarding a condition not listed please contact us on 0800 800 337 or email info@mda.org.nz

IMPROVING QUALITY OF LIFE


Bariatric Lux/Ultra Low


Vendlet V5S


Diagonal Toilet Lift


Ropox All-in-One Hoist

Bariatric Lux - The Bariatric Lux 4 section electric bed offers both functional design and excellent quality. The beech wooden paneled bed ends have been developed to offer the user an attractive domestic look . The extra sturdy frame allows a safe load of up to 318 kg. Operation via the hand control allows the occupant to select various positions as required. The bed offers two various heights ranging from 42 cm and 81.5 cm (LOW VERSION:25.9 cm-62.5 cm). The mattress platform also offers trendelenburg positions. Two further actuators beneath the platform allow full profiling capability of the backrest and knee break features.

Vendlet V5S/Speed Adjust - The New VENDLET V5S is an automatic patient turning system for moving and handling the bedridden client with limited resources. When the bars are raised the VENDLET V5S works as a side rail. The side rail function is tested and approved by TÜV. The speed adjustment makes it possible to reduce the speed of the bars to 75 percent or 50 percent of the normal speed. Each system is supplied with 2x slide sheets and 2x Turning sheets as standard.

Diagonal Toilet Lift - Toiletlift Diagonal is based on the natural movement of standing up, offering optimum support, with both feet firmly planted on the floor, during the 'stand up' phase. An important feature of this Toiletlift is the ability to adapt to the users specific height and weight. With no bar in front of the Toiletlift, the user is able to move their feet back to facilitate 'evacuation'. Ideal for users who have multiple sclerosis, Motor Neurone disease, post Polio, Hemaplegia, Strokes and other disabling conditions. The weight rating of the AEROLET Diagonal is 150 KG with a bariatric version rated at 250 KG also available. The toilet lift can also be fitted with a Bidet.

Ropox All-in-One Hoist - Our "All-in-One" active hoist can be changed from a standard hoist to a standing hoist without the use of any tools. As a Combined patient- and stand-up hoist the product provides excellent support and safety for the user and it improves the working area conditions for the helper when lifting the patient in different situations.

euromedical ltd
IMPROVING QUALITY OF LIFE
6/68 Dunlop Road, Napier, 4110
www.euromedical.co.nz | info@euromedical.co.nz