

# InTouch

KIA NOHO TATA

Inclusive | Inspiring | Informative

Winter 19 Issue 102

## California Dreaming

Great planning the  
key to achieving  
your goals

Leaping into  
the unknown

Why it's such a good move

How to hire  
support workers

To let you get on with living

Voicing our concerns

On the steps of Parliament



Muscular Dystrophy  
New Zealand



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New Zealand**

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We would also like to acknowledge our corporate sponsors:



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 with Trevor

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The organisation has another major year ahead as we approach our 60th anniversary as an Incorporated Society.

Our AGM on April 26 signalled the fact that another year had come to an end and simultaneously presented me with the opportunity to report to our members.

It has been an absolute pleasure to serve you as National Executive Chairperson following a period of change in the organisation where I stepped up into the chairperson role (from vice chairperson) part way through my two-year term.

And I am delighted to have been elected National Executive chairperson for a further two-year term. I give my word to do the best I can for all our members.

The National Council is Tristram Ingham, Vice Chairperson; Jan Daly; Joy Jenkin; Andrew Willetts; Brent Walker; Scott Laurenson; Scott Boyle; Mike Schneider (Northern); Bernadette Ingham (Central); Matthew Willetts (Southern). Canterbury Branch rep to be filled.

A significant achievement for the organisation was becoming recognised as a Disabled Persons Organisation (DPO) and officially joining the DPO Coalition in September. This role now offers us a seat at the table with Government, amplifying the voice of our members and contributing to a more responsive society for disabled New Zealanders.

I acknowledge former CEO Ronelle Baker and councillor Tristram Ingham for acting as the MDANZ representatives and facilitating our involvement with the DPO Coalition.

Our first group of bronze level Duke of Edinburgh "Dukies" are close to completing their awards. I thoroughly enjoyed being involved in the three-day adventurous journey which was trialled in Auckland as an urban-based journey to accommodate the different mobility needs of the group.

It was great to see this determined and positive bunch of young members come together to share new experiences. We will continue to develop and invest in this programme in the hope that some of the current Dukies will advance to the silver level award, and that the next set of bronze award level Dukies will be underway shortly.

The MDANZ Members Discretionary Fund continues to be a fantastic initiative and is making a positive difference to our members. This fund is something I fully support, as it is an awesome way to give a little back to our members.

The number of applications continues to grow with each round and although we would love to fund every application, it is impossible to do so. However in 2018 we received 60 applications and were able to fund 42 of those applications in some way.

Some of the things we funded were conference attendance, travel, hearing aids, funds toward an electric bike, a tablet, vaccinations, funds toward university fees and a key lockbox for support workers to safely enter a property.

We continue to invest in core services for our members including free counselling, information via *In Touch* and the website, as well as our fieldworker service.

The organisation has another major year ahead as we approach our 60th anniversary as an Incorporated Society. We do hope members and supporters will join with us in August when we celebrate our 60th Jubilee.

A handwritten signature in blue ink, appearing to read 'T. Jenkin'.

Trevor Jenkin  
National Executive Chairperson

## An alternative form of photo ID:

### The Kiwi Access Card


One difficulty in getting banking services for some disabled people can be trying to open a bank account without a driver licence or a passport.

In response to this a new "Kiwi Access Card" was launched earlier this year.

The photo ID card is a secure and reliable way of proving your age and identity. It can be used to access goods and services, such as making hire purchases, picking up prescriptions or opening a bank account.

The Kiwi Access Card replaces the 18+ Card which was a less comprehensive form of photo ID. It can be used as proof of age and identity throughout New Zealand and is available to both New Zealanders and foreign visitors.

The card has been designed to ensure it is compliant with the latest enhanced security features. It includes embossing and micro-text, as well as braille.

For more information about how to get a Kiwi Access Card see: [www.nzpost.co.nz/personal/realme-id-apply/kiwi-access-card](http://www.nzpost.co.nz/personal/realme-id-apply/kiwi-access-card) 



## You are invited

Here's two milestone events you won't want to miss.

We are incredibly excited to invite all our members and friends and families to our Freedom Beyond Limits, Neuromuscular Education and Advocacy Seminar then to get out your glamour style for the Muscular Dystrophy Association New Zealand Diamond Jubilee celebration.

Both events are taking place on Saturday August 3 at Waipuna Hotel and Conference Centre in Auckland's Mt Wellington.

The 2019 Neuromuscular Education & Advocacy Seminar offers an exciting programme aimed at improving the health and wellbeing and advocacy skills of individuals, families and whānau living with neuromuscular conditions, through research, education and collaborative practice.

Issues to be covered during the day include new treatments for neuromuscular conditions; living well and resilience and gratitude in response to sorrow. Our speakers include Rob Besecker, Dr Gina O'Grady, Assoc. Prof Richard Roxburgh, Julie Rope, Dr. Larry Stern and more.

The seminar will combine with a milestone event as the Muscular Dystrophy Association of New Zealand (MDANZ) celebrates its 60th Jubilee.

### **Freedom Beyond Limits: Neuromuscular Education and Advocacy Seminar**

**When:** Saturday 3rd August 2019,  
9.00am – 5.00pm

**Where:** Waipuna Hotel and  
Conference Centre - 58 Waipuna Rd,  
Mount Wellington, Auckland

**Price:** \$50.00 incl. GST for members  
& \$109.25 incl. GST for non-members/  
health professionals

### **MDANZ Diamond Jubilee**

**When:** Saturday 3rd August 2019,  
6.30pm – 10.00pm

**Where:** Waipuna Hotel and  
Conference Centre - 58 Waipuna Rd,  
Mount Wellington, Auckland

**Price:** \$45.00 incl. GST for members  
& \$86.25 incl. GST for non-members/  
health professionals

**Dress Code:** Formal

See [www.mda2019.org.nz](http://www.mda2019.org.nz)





## A cup of tea and a catch up with Samara Somerville-Petch

Each issue we introduce a MDANZ team member:

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

I have worked for the organisation since July last year, so around 10 months now. I'm currently the fundraising assistant which means organising our Annual Freedom Appeal and various other projects to help promote MDANZ.

### What qualifies as a great day at work for you?

I love seeing the work I'm doing really making a direct impact for our members whether it's getting members involved in our community or organising events so we can promote awareness of neuromuscular conditions. I love interacting directly with members and actively working to make them feel supported, encouraged and fulfilled.



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


I would love to provide more extensive mental health support for our members.

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

I can never go past a good hummus and crackers combo.

### What are you passionate about?

I'm really passionate about doing meaningful work that has tangible results. Also classical music and opera is a life-long passion of mine.


If you ever want to contact me personally on [samara@mda.org](mailto:samara@mda.org) I'm always here to help or listen. 

## I Choose carer subsidy

In our summer 2018 edition of *InTouch* we discussed the "I Choose" carer subsidy which was due to be available from December 1.

The roll out of this new initiative, which provides more flexibility for

people receiving funding for respite care, has been put on hold until more information has been gathered to ensure it is fair and equitable to all.

The Ministry of Health will issue an update once the roll out is confirmed and we will keep you updated via Facebook, *InTouch* and email. 




## DPA Youth Project Facebook

DPA is currently undertaking a Youth Project led by one of our members, Joe Boon, to connect and engage with disabled young people.

As part of the project Joe has set up a closed Facebook group (one in which the group name and member list are public, but the discussion can only be seen by members of the group).

Joe is inviting disabled young people to join the group. He says: "There is no precise age limit, but if you self-describe as young or are between 17 and 35, please consider joining our Facebook group and get some discussion going."

The link to join is: <https://www.facebook.com/groups/774758249574922/> 

## CSC member discount card

Your 2019 member discount card is now available. Do make use of it.

To receive great discounts instore, at the checkout, show the supplier account number on the card. If online enter the promo code listed when ordering. If you do not receive a discount you are expecting, please ask to speak to the manager. The staff may not be aware of the discounts offered.

This year's suppliers include:

**AwaytoGo:** Excellent gift vouchers that are purchased online. The AwaytoGo website on your card is no longer valid and the new one is <http://bit.ly/CSCATG>.

**Beaurepairs Tyres:** Up to 15

percent off Dunlop, Goodyear, BF Goodrich and Michelin tyres.

**Capes Medical:** More than 10,000 medical items available. All products discounted by seven percent and include GST. Also offers competitive discounts for incontinence products and gloves.

**Hertz Hire:** Up to 35 percent discount depending on the type of vehicle. Also available in Australia.

**OfficeMax:** Office stationery, computer consumables, technology, furniture, safety gear and more. Up to 20 percent discount.

**Pit Stop:** Ten percent discount on a variety of products and services such as repairs, safety checks, WOFs.

**PlaceMakers:** All building supplies, equipment and advice. Discount varies.

**RepcO:** NZ's largest supplier of automotive and marine parts and accessories. Ten percent discount on retail pricing on most items (excluding specials and promotions).

To get your 2019 member discount card contact your fieldworker or national office on 0800 800 337 or [info@mda.org.nz](mailto:info@mda.org.nz). 📞

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## A global registry for SMA

Neuromuscular Research New Zealand (NRNZ) continues as the principal funder of the New Zealand Neuromuscular Disease (NZ NMD) Registry. The registry is credited with facilitating almost all neuromuscular related research in New Zealand and has achieved enrolments of more than 1,200 people with neuromuscular conditions.

The curator of the NZ NMD Registry has a lead role with an international group, TREAT NMD, developing a global patient registry for patients with spinal muscular atrophy.

In addition to the registry, research funded by NRNZ in 2018 included:

- An international collaboration by Dr Luciana Pelosi investigating nerve ultrasound in spinocerebellar ataxia.
- Dr Silmara Gusso's study looking into vibration training for children and young people with congenital myopathies. <sup>N</sup>

## Access and safety of non-invasive ventilation

A study exploring access and safety of non-invasive ventilation for neuromuscular disorders led by Alister Neil was funded by the Neuromuscular Research New Zealand in 2017 and completed

in 2018. Study findings will be presented to health funders and the clinical community, with the hope of improving clinical services and health outcomes for New Zealanders with neuromuscular conditions. <sup>N</sup>

## Our funding

In 2018 the trust itself received grants from Richdale Charitable Trust, MDANZ, and the Stern Foundation to fund the research projects in 2019.

These grants made up 86.4 percent of Neuromuscular Research New Zealand's income while donations contributed eight percent and interest received contributed 5.6 percent of our income. Meanwhile, Neuromuscular Research New Zealand annual grants funding round held in September last year received five applications, and following review by the trustees, four grants were awarded funds and the fifth was declined. <sup>N</sup>

## Together in SMA

The 2019 Australia & New Zealand Together in SMA Healthcare Professionals Forum was held in April in Melbourne. Around 80 healthcare professionals including Sharron Meadows, Starship Paediatric Neuromuscular nurse and Miriam Rodrigues came together to share and learn all things SMA.

The NZ NMD Registry, of which MDANZ is the primary sponsor, was showcased at the forum as development of a global network of SMA registries continues.

Patient registries are essential for facilitating research, understanding the natural history of rare conditions, facilitating the development of improved standards of care, and monitoring the effects of therapeutic interventions. <sup>N</sup>







Muscular Dystrophy Association NZ

## Freedom Campaign 2019

Throughout September we'll be highlighting the triumphs and challenges experienced by our members. The month-long campaign will be kick started with our National Appeal Day on Friday, September 6.

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What does Freedom mean to you? As we fast approach our annual Freedom appeal we're keen to start this conversation to further our mission of promoting freedom of choice and a responsive society.

Our Freedom Campaign is all about raising both awareness of neuromuscular conditions and funds for local services. This is a time to celebrate and emphasise that everyone should have an opportunity to experience freedom beyond limits. Neuromuscular conditions can present in people of all ages and backgrounds. Each of our members has lived experience of neuromuscular conditions and we'll be showcasing their wonderful achievements, each having a unique journey with different triumphs and challenges along the way.

The 2019 Freedom Campaign comprises various events around the country that will serve a dual purpose of raising awareness about neuromuscular conditions as well as much needed funds for our organisation.

Friday 6 September is our National

Appeal Day and we encourage you to celebrate and support us in any way you can – you might like to host a morning tea with work colleagues or friends or organise a mufti day at your local school.

Watch out for us on social media leading up to this day and remember a 'like' or 'share' can go a long way. Come along to our fundraising events and support our community.

We'll be celebrating our members by sharing their stories on social media further raising awareness of this amazing cause.

Ways to get involved this Freedom Appeal:

- 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.
- Hold a morning tea for Muscular Dystrophy.
- Join the schools around the country already holding mufti days.





## Comfort Cough available for purchase or rental

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[www.breathingandmedical.co.nz](http://www.breathingandmedical.co.nz)

Phone: 0800 335 333

## Freedom NEWS

We have always been delighted by the amazing member events and efforts that have been seen throughout the country.

Get in contact with your local branch or fieldworker to express your interest. Alternatively, you're welcome to register interest through our website [www.mda.co.nz](http://www.mda.co.nz). 



## Freedom diary dates

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Put these dates in your diary for our upcoming Freedom Appeal. It's going to be the best one yet!

### **7th Sept – DMD Awareness Day**

#### **7th Sept – Southern Community BBQ at Dunedin Bunnings**

Come along and grab a snack while supporting Muscular Dystrophy at the same time.

#### **7th Sept – Northern Gala Day in Central Auckland**

This Gala Day is filled with fun for all the family. Members and the public alike can enjoy the day with wheelchair racing and gumboot competitions.

#### **16th Sept – Southern Quiz Night in conjunction with Parent to Parent at Northstar Oamaru**

How good is your general knowledge? Enjoy a laugh and a drink in support of two amazing organisations.

#### **22nd Sept – Northern High Tea in Central Auckland**

Calling all ladies to a Sunday afternoon gathering with small sandwiches, scones and tea of course.

#### **24th Sept – Southern Quiz Night at the Kensington in Dunedin**


Another test of your general knowledge. Maybe you'll be better than the first time or just coming back for more.

#### **27th Sept – Northern Member Morning Tea in Central Auckland**

Members come one, come all for a great informal catch up. We would love to see you there.

#### **29th Sept – Limb Girdle Awareness Day**

#### **29th Sept – Northern Limb Girdle Walk in Whangarei**

The Basin in Whangarei is a perfect place to raise awareness for the Limb Girdle condition. Join us in supporting families with this condition. 

# Hiring support workers to let you get on with living

## *Choose the option that fits you best*

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Hiring a support worker may not be as straight forward as you might imagine. Natalie Brunzel outlines what she wishes she had known before hiring her own supports.

When you first apply for support funding, whether it's IF, EGL or any other type, there is just one thought that pushes you through the hours of assessment and the whirlwind of paperwork – you need the support to get on with living.

So, you have received the email confirming your funding and are now able to go ahead and choose your host and how to implement the supports you need. All of this sounds straightforward, however there are things I wish I had been told about hiring my own supports.

Many people think that when we speak about hiring support workers we are automatically talking about becoming an employer. Do you know what the options are and what responsibilities you have? I sure didn't!

I didn't know that there were any other options than employing support workers myself, effectively forcing me to become a business and having to learn about all the regulations that come with that.

I began employing people

and I have to say my host made it very easy, providing me with all the information I needed to move forward and hire the help I needed. However, there was one thing that always bothered me about employing supports. Many of the individuals that worked with me felt like more than employees and being their 'boss' didn't sit well with me at times.

I asked myself whether there was a way to hire people

that took away the power dynamic automatically created through being a boss to an employee. It was only through discussions with friends and other members within the disability community that I discovered the various ways people hired supports and balanced the power dynamic.

What I have learnt is that there are three ways in which you can hire your support workers: employing, contracting, or as IR56 employees.

### Employing

You are required to become an employer. You will need to know whether the individual will have a permanent or casual agreement. This is a formalised agreement and you will need to ensure you deduct PAYE and provide the necessary holiday and sick leave. It is also up to you to pay for ACC and to manage all the other regulations that govern employees.

The benefit of having this type of agreement is that you can stipulate leave-notice periods and you can provide regular employment hours.

There is software that can support you – hosts such as Manawanui have software that can support you to run this effectively, using online portals which enable you to complete all the paperwork online, including providing you with up-to-date information on your statements.

There are plenty of other options on the market to choose from, including Thankyou Payroll, Ace Payroll, Crystal Payroll, IMS Payroll and many more. Many payroll services offer a free trial period, so make use of this to see whether this option really works for you.

All hosts have coaches that can help to ensure that you



Natalie Brunzel





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*It was only through discussions with friends and other members within the disability community that I discovered the various ways people hired supports and balanced the power dynamic.*

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have all the resources, and they offer payroll services that enable the management to be easy. There is often a cost involved with having these services. If you decide to go this way, check the extra costs that each host offers, as they differ. One host might be better value depending on the size of your package(s) and the types of support you are receiving.

One drawback that I found was that it can be hard to track your budget as you need to ensure you know how much is being put aside for holidays and have money set aside to pay for another worker to cover the holidays. And as mentioned previously the power dynamics can be difficult to manage.

You also need to pay to advertise for new support workers – advertising is another aspect which seems to be fraught, but we won't cover it in this in this article.

## Contracting

This is when your worker is an independent contractor, meaning effectively they are their own boss. You have a contractual agreement that can be cancelled at any time. You clearly outline the tasks and times.

The benefits of this arrangement are that the person working for you can claim back their expenses, leaving more money in their hand. You will normally pay them a higher pay rate to cover holiday, sick and bereavement leave, which they are responsible for.

Mycare is a contracting platform that provides you with an online service for finding and hiring support workers safely and securely with everything done online. This means there are no timesheets or other paperwork to deal with.

It also has technology that enables you to have 'circles of support', giving support workers the ability to leave notes for one another online, and there is no payroll cost. This means more of your funding can go straight to the worker.

I have found that having an online service works well for me because I don't have to manually fill out timesheets, making it much easier to manage when I am travelling or just busy with life.

## IR56

An IR56 taxpayer is an employee that is required to pay their own PAYE through an IR56 form. This includes part-time, private domestic workers who are employed to work in their employer's home but not in work related directly to the employer's business work.

This is an option that many of my friends have opted for as it enables them to still have the benefits of employing people while giving workers the ability to be their own boss.

There are of course, pros and cons to any way of organising your supports. That is why it is important to be well-informed and know what would work for you. Make sure you choose the one that is going to support you best. Remember technology is your friend, so familiarise yourself with the various apps that might make your life easier. It is your life and you should be able to choose the option that fits you best.

# Finding the Freedom Chair advantage

If you find walking difficult then the Freedom Chair could help. It's great for indoors, outdoors and when you want to travel. The Freedom Chair is one of the lightest and most versatile alternatives to a mobility scooter and it's a great choice for all round convenience and performance.

Driving the Freedom Chair is easy and fun. Simply press the power button on the controller, set the speed and use the joystick to move around. Low speed is perfect for around the house or in buildings. Manoeuvring in passageways and through doors is easy and this also makes it safe for beginners to learn with confidence. Medium speed is perfect for keeping pace with sidewalk traffic or a friend on foot. In high speed the Freedom Chair will take you up to 6.5 kilometres per hour.

Indoors or out, on trains, planes, or automobiles, when it's time to go anywhere, there's no better travel companion than a Freedom Chair.

**Robert's story** "I've been an MS sufferer for many years and have pretty much lost all mobility on my right side. This has been severely restrictive. I already have a large power chair and I've been using this chair for both inside the house and for outdoor trips which has certainly made my life more bearable.

"However, because of its size and weight, loading this chair into our small SUV by joining three pieces of aluminium to make a ramp is time consuming and a little difficult for my wife, Sharon. Once we arrived at our destination we needed to find parking where we had the space to install the ramps, lower the chair to the ground and do this without impacting on those around us. I then discovered the Freedom Chair. Paul from Montec Mobility sourced the chair and trained me how to use it. I'm now FREE.

"Sharon and I can go out together with ease as it only takes a few seconds for her to load the Freedom Chair into the car. Parking is unrestricted as there is no need for a ramp.

"The Freedom Chair has given me my life back and the best thing I can say about it is: It's Life Changing." – *abridged.*



## A Versatile Power Chair



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## California dreaming

*Great planning, it seems, is the key to reaching your goals*

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A long-held goal to visit the Golden Gate Bridge yielded far better things than Hamish Taylor had hoped for.

Hamilton-based Hamish Taylor has always dreamed of going to the United States. The lure of the Golden Gate Bridge, the buzz of Las Vegas, the fun of Los Angeles and the grandeur of the Grand Canyon were all on his radar. And two years ago, Hamish and his family celebrated his 21st birthday by visiting all the spots Hamish had dreamed about.

In fact, Hamish is so enamoured of all things American his father, Steve, quips he would probably go and live there if he could.

Hamish tells *InTouch* his first goal was to see the Golden Gate Bridge in San Francisco and as they began planning their two-week family trip, the goals broadened considerably.

Steve says as they began to map out the trip, Hamish pointed out that once they were in San Francisco, LA was

only five hours drive away; and from there Las Vegas was just four hours down the road and then it wasn't that much further to the Grand Canyon.

"I said he had to stop there. I wasn't driving the whole of the United States," says Steve.

Hamish's mother Sharyn and sister Lucy were on board for the trip and while Hamish's younger brother Austin couldn't come, they intended to take him at a later date.

Both Hamish and Austin have Duchenne muscular dystrophy (DMD) and as a result Hamish has severe scoliosis in his back which makes it hard to sit for long periods.

That could have been a problem on the 12-hour flight, but Hamish says while he was wondering how that was going to work, the family found the Air New Zealand SkyCouch option, where a row of seats fold down, which meant that Hamish could lie down.



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*Hamish's first goal was to see the Golden Gate Bridge in San Francisco and as they began planning their two-week trip, the goals broadened considerably.*

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The family was also able, through a contact of Steve's, to visit Air New Zealand's training area and look at the seating configurations before they booked the tickets. The MDANZ Discretionary Fund helped them pay for the extra seats Hamish needed.

The highlight for Hamish was the Golden Gate Bridge in San Francisco although he was impressed too with LA and Las Vegas. It was very hot in both places while San Francisco was a better climate for walking around.

They also had an accessible rental van with good air conditioning. Hamish says the hotels were very good and more advanced in catering for wheelchair access with better space in the rooms than in New Zealand. Transport was also more advanced, with the shuttle buses from airport to hotels being wheelchair friendly.

The trip, he says was everything he expected. He was taken aback by the size of the place and how big everything was, including the food servings.

And they didn't leave many stones unturned on the trip – in LA they went to Hollywood, Universal Studios and Disneyland. "There was just heaps to do."

In truth, he says, he could have gone for another

two weeks.

For Austin, who couldn't accompany the family to the US, but had been planning his own US trip to celebrate his 21st later this year, medical complications meant the family had to rethink. And he has since had his own adventure travelling around the lower South Island with his

parents and sister and getting across on the ferry to the wild beauty of Stewart Island.

As for his next trip Hamish is keen to see the V8s race at Bathurst in Australia as both brothers are interested in the big cars. A cousin on their mother's side is the well-known racing driver Scott McLaughlin who catches up with the family when he is in town.

### Tips for parents wanting to travel

Steve Taylor says that hiring a wheelchair friendly vehicle in the US is easier than here, mainly because of the population base. He found three or four companies that specialised in accessible vehicles.

That said he did find the customer service less than optimal and when rental companies did not get back to him via email he began getting up at 4 a.m. to phone them and found this far more effective.

The biggest part of the journey was all the planning. "Once we got there it was pretty easy."

Hamish and his dad spent a lot of time preparing. While they initially used the hotel aggregation sites like Trivago when they found a hotel that looked suitable, they contacted them directly. And once they found a winning hotel, they used that chain in all the centres they visited.

While many accessible rooms in New Zealand are only for the person in the chair and a support person, in the States they had full family rooms.

His advice to other parents and care givers planning an overseas trip is that it's all in the planning.

"Everything is there, you just have to find it. You have to be prepared to give the hotel or rental companies a call and don't be afraid to tell them what your requirements are. They are better set up than we are here."

As to getting around the country Steve says to be aware that different US States have different rules around mobility parking. In some States you can use your NZ mobility card, but in California they went to a local authority and showed them the NZ card before they issued one for California.

Steve is well versed in mobility as he has his own business, Tailored Access, which undertakes mobility audits, and he is also a BE Accessible coach.



Austin and his sister Lucy at Invercargill's Transport Museum.

# Living life on the edge

## *Melanie Eggers lives her life to the max*

Sometimes overcoming great challenges in life can mean setting some very, very high goals for yourself. And that is what sky-dive and bungy jump lover Melanie Eggers has done over and over again.

Melanie Eggers likes to live life on the edge and encourages others to do so too. In her view: "You have got to take your chances and if you want to do something, do it. Just live life to the max and do what you like."

And she follows her own advice pretty closely.

Palmerston North based Melanie undertook her first sky dive five years ago, jumping 13,000 ft at Auckland's Helensville air base, all on the same day she saw the legendary Bruce Springsteen in concert.

Her latest sky dive, this time from 20,000 feet, was late last year and again on the same day she saw a major international act perform in Auckland. This time it was Def Leppard.



Top: Melanie, on her recent wedding day, with husband Dave and 13-year-old stepson, Clint. Above: Melanie after her brain surgery.



High above Auckland city on the Sky Walk. Nothing is too high for Melanie.

"Concerts and sky diving are what I do," she says.

On top of this Melanie has completed nine bungy jumps including jumps on the Gold Coast in Australia, Auckland Harbour Bridge and the Auckland Sky Tower twice. She has also had fun on bridge swings.

Essentially, she says, she loves heights and in her book, jumping backwards on a bungy is the best way to go.

"It is more fun seeing where you have been, than where you are going."

And Melanie is not unused to publicity as she appeared on the cover of *InTouch* magazine

*"It is more fun seeing where you have been, than where you are going."*

in our Summer 2016 issue where she told the story of meeting her birth father Simon.

Melanie has glycogen storage disease, which is an inherited condition that impairs the body's ability to breakdown a complex sugar called glycogen in the body's cells. In researching the condition, she discovered another person living in Palmerston North with the disease. Melanie and Simon started chatting online and over time it transpired they were father and daughter. Their story also featured in *That's Life* magazine.

They first met four years ago and they have since become close.

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*"As long as I can walk or jump, I will keep doing it. Having been invalided all my teenage years and in my 20s, I am living life now."*

---

Today Melanie credits a major brain operation for taking away her fear heights and completely changing her life.

In 2005, when she was 26, Melanie had a right temporal lobe lobectomy, where an area the size of a matchbox on her right side was removed, to help the extreme epilepsy she had been suffering from since she was 13.

She recalls she had been on a waiting list for a year and finally got the call on December 8, 2005.

Until that point the epilepsy had meant "I couldn't even cross the street on my own."

Her seizures have not only gone but she now drives, holds down a full-time job at a pharmaceutical company and, as *InTouch* spoke to her, was planning her wedding in her garden, which was to take place the day before her 40th birthday. Her birth father Simon was giving her away on her wedding day.

"I have such an amazing life now, from what I had."

Melanie and her new husband Dave were heading to Queenstown for their honeymoon "so I can bungy jump and maybe sky dive as well".

Her husband, Dave, who she works with, won't be doing so, as he is scared of heights, she says.



Sky diving: Melanie says it is an amazing feeling to free-fall at 200 kilometres an hour for 90 seconds.

But it will never be high enough for Melanie. "If I can see the ground it is not high enough."

As to why she loves the sky diving so much, she says it is an amazing feeling as you free-fall at 200 kilometres an hour for 90 seconds.

And she is adamant she is going back for more.

"As long as I can walk or jump, I will keep doing it. Having been invalided all my teenage years and in my 20s, I am living life now."

So, should others do it? "Absolutely," she says, "lots of people take everything for granted."

But she admits that getting married is a completely different thrill. "It is really cool getting married again and becoming a step mum."



Muscular Dystrophy  
New Zealand



*Beyond DNA*  
Stories of Challenge and Triumph

# Beyond DNA

is a moving and inspiring collection of personal stories which reflect on life lessons learned by those with lived experience of a neuromuscular condition.

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# Leaping into the great unknown

*You can learn and gain so much if you do*

---

Tackling a three-year degree, in an environment he wasn't used to, with a whole new group of people and circumstances he hadn't encountered before was very much out of Ben Yellowlees comfort zone. He says, now, he would tell himself: You got this.

I often wonder what I would say to myself if I was able to travel back in time and talk to myself but at a much younger age. What lessons I would give or advice I'd share. It would probably vary from age to age but given the opportunity to speak to myself, four years ago, less than a month away from starting an intense three-year degree at university, what I would say would be simple. I would tell myself this:

You got this.

You see, I was a lot different four years ago. I've grown up an incredible amount since then and have evolved so much as a person. I credit a lot of that change to my experience going to university, and I have it to thank partly for me being who I am today.

So let me tell you a little bit about my journey from an anxiety-ridden teen afraid to meet new people and try new things, to a more open minded, confident young



Ben credits the immense motivation and encouragement he received from his parents as part of the reason he decided to go to university.

adult and talented graphic designer, as well as some of the struggles I overcame along the way as an SMA sufferer.

To say that tackling a three-year degree, in an environment I'm not at all used to, with a whole new group of people I don't know, and circumstances I haven't encountered was out of my comfort zone as a 19-year-old leaving the familiarity of high school, would be a gargantuan understatement.

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*One of the biggest things was going through my final year and a half with no support worker with me.*

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Then you have the added difficulties we as disabled people face on a daily basis. As someone who loves their comfort zone very much, probably a bit too much, I was extremely hesitant about this leap into the dreaded unknown.

This begs the question, why did I do it? The obvious answer would be my passion for design and my capabilities in the subject. But, perhaps a just as important reason, was the immense motivation and encouragement I received from my parents.

They knew that it would be a challenge for me but also that it would ultimately be absolutely worth it. They saw the value in pursuing a degree, no matter how many obstacles and unknowns I would face, and in turn allowed me to see the value in it as well.

They were right, it was totally worth it. But it wasn't always straightforward or easy, and there were times where I wanted nothing more than to give up.

Part of the course work required to get my Bachelor of Design was opening yourself up to a lot of honest critique, making lengthy presentations in front of many people, getting involved in a lot of group activities and adapting to the fact that I would have to do things I was uncomfortable doing, over and over and over. Even well

into my third year, I would get nervous stomach aches on my way to class knowing I had a 10-minute presentation that morning to showcase my latest work.

There were times I genuinely questioned whether I had the skill or talent to even belong there and create work that was good enough, constantly comparing myself to the other people in my class.

One of the biggest things though was going through my final year and a half with no support worker with me. This altered so much of my uni life and how I dealt with obstacles and it was something I never imagined occurring.

However, thanks to the helpfulness and generosity of my classmates and lecturers, the friendly hours of my major and my own self-belief and determination to succeed and make my family proud, I got through it relatively easily. As I did with every other challenge one way or another that I faced throughout those three years at AUT South Campus.

What I'm trying to say is life is filled with these periods of uncertainty, adversity, discomfort and apprehension, that we as human beings naturally tend to veer away from and disassociate ourselves from in order to remain in a mental state of contentment and convenience.

It's hard not to believe this feeling of complacency can be heightened when you have a disability. But what we should do when we encounter these scenarios is embrace them and not shy away from them.

You can learn and gain so much if you just drag yourself up the hill long enough to witness the view at the top.

I'm 23-years-old now and, while I still have a ways to go and am by no means the altogether confident, highly successful, faultless version of myself I wish I could be, I am undoubtedly proud of my progress and how much better and more complete I am now compared to my former self.

I'm more confident in my ability as a designer as well as a communicator and in my interactions with others. My knowledge of, not only design, but the world has expanded and the life skills I've learnt have been invaluable.

Moving forward in life with my freelance design business Benji Design, I am self assured that my decision to study was not only valuable but the right decision for me as a young man and a graphic designer. And I take pride in the fact I did it.



Ben is moving forward in life with his freelance design business Benji Design.

---

*What we should do when we encounter these scenarios is embrace them and not shy away from them. You can learn and gain so much if you just drag yourself up the hill long enough to witness the view at the top.*

---

I feel prepared and confident to continue building my brand, finding work and putting my designs into the world.

Even though at times it may be slow and steady. I look forward to what the future holds for me and my design work.

# Medicines for rare disorders

## *How PHARMAC funds these medicines*

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Miriam Rodrigues explains how PHARMAC is funding improved access to medicines for rare disorders.

PHARMAC is our government agency that decides which pharmaceuticals to publicly fund in New Zealand. While PHARMAC's utilitarian approach of providing the greatest good for the greatest number, within its budget set by the Ministry of Health, works well for most of us, most of the time, it didn't work so well for those of us with rare disorders especially when the medicines available are expensive.

To address this, in 2014, PHARMAC piloted a commercial process aimed at improving access to medicines for rare disorders.

A lack of competition was seen as a barrier to getting better access to medicines for rare disorders and so up to \$5 million per year for five years was made available to fund a contestable pilot.

This resulted in 10 medicines being approved for listing on the Pharmaceutical Schedule through this process, including Myozyme for infants born with Pompe disease.

An external evaluation of the pilot concluded that better commercial proposals were received, and funded access to treatments for rare disorders improved.

In late 2017, PHARMAC announced that it was introducing a set of dedicated features for considering medicines for rare disorders, so that existing processes could be well-utilised.

Rare disorders were defined as "a clinically defined disorder affecting an identifiable and measurable patient population with a prevalence of less than 1:50,000 in New Zealand".

And the special features introduced included:

- A standing Pharmacology and Therapeutics Advisory Committee (PTAC) expert subcommittee for rare disorders.
- Regularly calling for rare disorder funding applications.
- Undertaking dedicated pre-engagement with new, and existing, suppliers prior to each call for funding applications.

- Formally adopting adjusted policy settings for rare disorders treatments.
- Regularly reviewing the portfolio of medicines for rare disorders and progressing good or reasonable opportunities for investment through routine process. Additionally, if a portfolio of rare disorders investments offered an opportunity to better obtain health gain, then PHARMAC would run another round of contestable funding or develop an alternate commercial approach, dependent on circumstances.

PHARMAC's Rare Disorders Subcommittee of the Pharmacology and Therapeutics Advisory Committee (PTAC) was established last year to provide advice on funding applications for medicines for rare disorders and had its first meeting in November 2018, where it considered nine medicines for 12 rare disorders.

Five of these received a recommendation for funding and one, Spinraza for spinal muscular atrophy, received a recommendation that a decision be deferred pending further evidence.

PHARMAC has initiated its second call for funding of medicines for rare disorders with the next Rare Disorders Subcommittee meeting scheduled for 24 September 2019. The subcommittee will consider new funding applications from suppliers or clinicians and also review existing funded medicines for rare disorders, including medicines considered via the Named Patient Pharmaceutical Assessment (NPPA) process for individual patients.



# Making our voices heard at Parliament

MDA members petitioning Parliament

Members of MDANZ and supporters were part of two astounding events at Parliament in early May where crowds gathered calling for PHARMAC to fund better access to treatments for a variety of disorders.

On May 1 more than 100 people gathered in front of Parliament to present a petition calling for PHARMAC to fund better access to treatment for people with Spinal Muscular Atrophy, of which there are less than 100 in New Zealand.

Fiona Tolich, an SMA advocate, said the petition, which was signed by 15,000 people, was presented to the Deputy Leader of the National Party, Paula Bennett. From there it goes before the Health Select Committee.

On May 7, Allyson, Christine and Freda joined Patient Voice Aotearoa at Parliament. This time it was Labour list MP Kieran McAnulty who will present their petition to Parliament urging changes to the PHARMAC model so more can access treatment.



Above: (Left to right) Allyson Locke, Christine Turner, Freda Evans, and Ruiha Evans. Top Right: Some of the crowd gathered in front of Parliament on May 1. Middle: Koen Gellatly (brother to Lincoln Woodmass), Fiona Tolich, Kuldeep Singh, Amber Collins (Dylan's mother) and Dylan. Bottom right: Members of Parliament and Patient Voice Aotearoa on May 7.

## Respite in New Zealand: We must do better

Carers are not being sufficiently supported in their caring role and this is leading to carer breakdown and situations where people can't cope anymore, a new paper has found.

A paper was commissioned by the New Zealand Carers Alliance late last year outlining ideas and actions to develop a fairer and more sustainable respite system for New Zealand.

It has been recognised that many parts of the respite system in New Zealand are broken and need fixing and therefore this paper focused on respite and not the suite of associated services which may influence the need for respite services.

The paper did not dwell on the problems with respite but focused on action and what is needed now to support improvement.

Respite is the chance for the carer and person being cared for to take a proper break. The aim of the paper was to promote an informed debate that will lead to action and improvement.

Below is a summary of this paper.

Most New Zealanders care for someone at some point in their lives. Whether that be a new born, an elderly parent or someone with a disability.

Sometimes the level of care provided goes over and above what is considered 'normal' and places considerable and, at times, unmanageable expectations on carers.

It changes relationships, causes stress, loneliness, lost income and lost sleep. At times, carers are unable to focus on themselves and in these circumstances need respite.



In the 2013 census, 430,000 New Zealanders identified as carers. This is equivalent to 10 percent of the population.

The report notes that: "Every year New Zealanders provide an estimated \$7 billion to \$17 billion of unpaid care. Imagine if everyone stopped providing this care. Imagine the massive financial and social impacts for the country."

Of all these carers, 31 percent to 61 percent of New Zealand respite users are able to use their full respite allocation.

### Who are they looking after?

There are approximately 1.1 million people living with disabilities in New Zealand. A survey found that 33

percent of disabled adults in private homes in New Zealand needed regular assistance for personal or domestic tasks.

New Zealand's population is getting older and the number of people living with dementia is also increasing, meaning more people will need extra care, and be unable to provide care to anyone else.

### Who funds what?

Respite services are funded through:

- District Health Boards for aged care, mental health and long-term chronic conditions.
- Ministry of Health for DSS.
- ACC for carers who support people who have had an accident.



Contracts may be long term or short-term depending on the situation. A Needs Assessment and Service Co-ordination (NASC) centre assesses entitlements.

## What's broken that needs fixing?

Carers are not being sufficiently supported in their caring role. This is leading to carer breakdown and situations where people can't cope anymore.

Respite is one critical area where we can support carers. However, services are currently not meeting people's needs, are not universally available and aren't adequate.

Currently there is the Carer Support Subsidy (changing to "I Choose") and Paid Family Carers.

Carers are saying that quality day services, or short break residential services are not readily available for the people who need them.

Service coverage across the country is patchy, with some areas

having better access than others. This is not due to funding but due to DHB priorities and focus.

For the services that do exist, there is limited quality assurance and monitoring. Respite services are not organised around people but around funding.

The current set up forces people to work around the system instead of the system working for them. Although pockets of excellence exist, they are not widely adopted. Overall, services exist in dysfunctional silos.

## Why we need to invest in respite

The population is ageing, and our services are not prepared. We know that when caring situations breakdown, the costs can be significant for urgent response services.

The annual health budget is \$15.6 billion, social security and welfare is \$30.6 billion and education \$14.1 billion. The value of care provided by informal carers is estimated to be

between \$7 billion and \$17 billion.

Informal carers provide huge amounts of value to New Zealand through their caring role and their contribution needs to be recognised.

Each week, informal carers in New Zealand spend an average of 24-36 hours caring for someone.

We need to change this conversation away from seeing carers as a cost, to seeing them as an area to invest in. We need to invest in respite to ensure carers are able to carry out their role without excess stress.

Investment does not always mean money. It means acknowledgement, information, and access to support if needed. It means recognition that caring for someone with needs that are over and above what others determine to be normal can, at times, place unmanageable expectations on carers.

It also means thinking more deeply about the consequences of not investing and seeing the implications over time.

Respite services should be developed to ensure that carers are supported and receive the help they need to continue caring.

## Action is needed to improve the system

We need to think about, and create, a coherent carer system, which has relevance across conditions, funders and locations. Effective respite is a key part of this system.

The four keys areas identified are: (1) carers and people they care for, (2) funders and commissioners, (3) providers and (4) the system of respite.





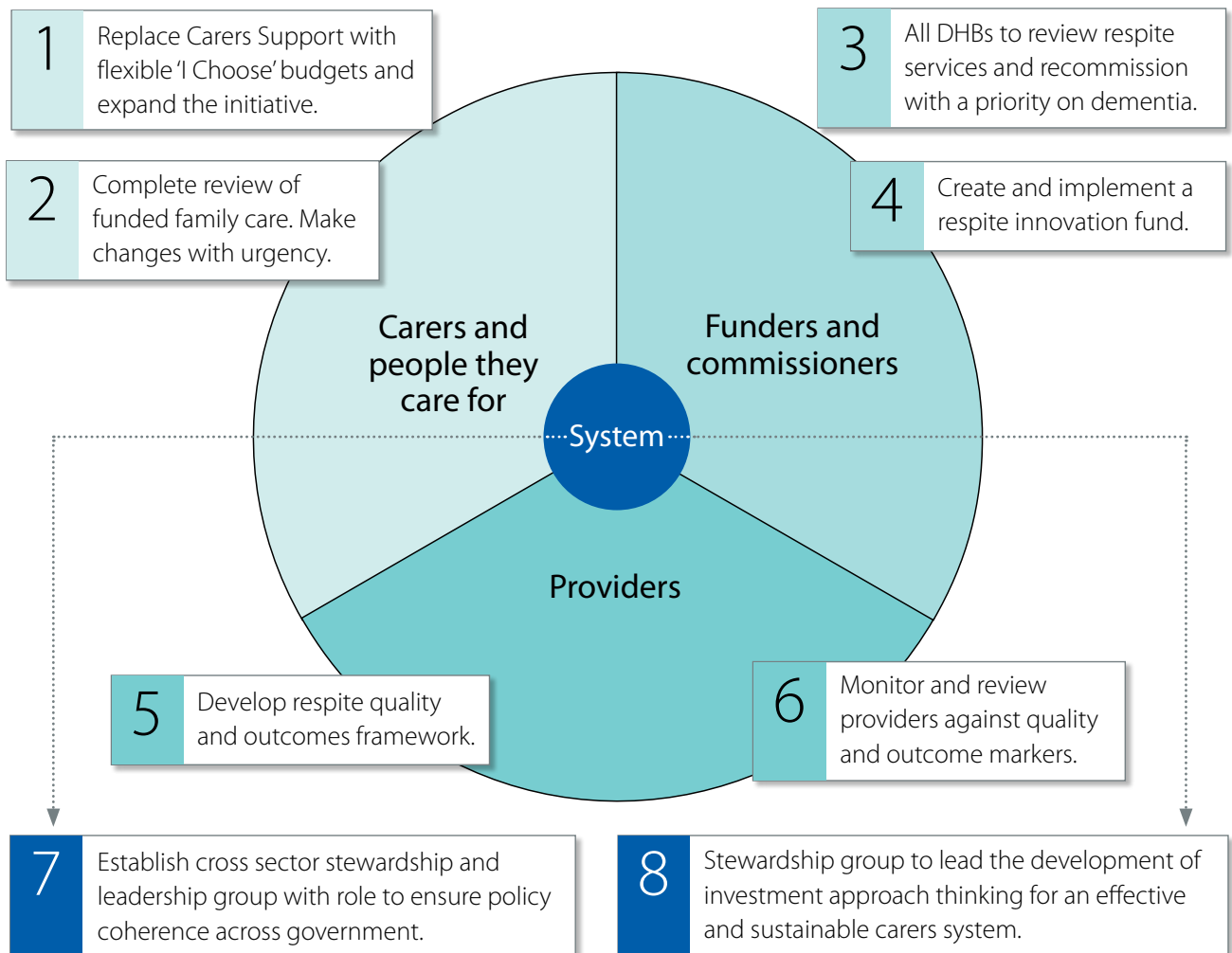


Figure 1: Focused set of actions.

Eight core actions have been proposed to improve respite services as described in the diagram.

An initial area of activity for the stewardship and leadership group should be to develop guidance for funders and commissioners on taking an investment approach for respite for all ages and populations. This needs to provide the rationale for the approach and practical guidance on how to target investment in carers.

### Start here....

The respite system is in crisis and action must be taken now. This paper suggests the following three actions be prioritised for immediate action:

1. All DHBs to review respite services and recommitment, with a priority on dementia.
2. Develop a respite quality and outcomes framework.
3. Establish cross-sector stewardship and leadership group with role

to ensure policy coherence across government.

These three actions can be started immediately and will kick start the changes that are needed to improve the respite system. <sup>R</sup>

*This paper was compiled and written by Synergia for the New Zealand Carer's Alliance. If you would like a full copy please contact national office on [info@mda.org.nz](mailto:info@mda.org.nz) or 0800 800 337.*



## A new potential treatment for myotonic dystrophy

A small molecule drug.

AMO Therapeutics is in clinical testing of a small molecule drug AMO-02 for treatment of congenital and childhood onset myotonic dystrophy and in particular the central nervous system issues.

To date, the large majority of therapeutic development efforts in myotonic dystrophy have targeted skeletal muscle.

The ease of accessing and evaluating splicing events in small muscle biopsies provides a drug developer with comparatively easy to obtain insights into whether the drug is working or not.

Assessment of the central nervous system in children and adults with myotonic dystrophy is more complex — the hurdles here include identifying appropriate drug targets and targetable CNS phenotypes and determining how interventional trials can best demonstrate the clinical effectiveness of drugs.

In efforts to establish sensitive and effective CNS outcome measures for congenital myotonic dystrophy, AMO Therapeutics has developed the Clinician-Completed Congenital Myotonic Dystrophy Type 1 Rating Scale (CDM1-RS).

The new scale builds upon prior efforts on clinical rating scales by DM researchers, Drs. Chad Heatwole and Nicholas Johnson.

The rating scale is currently being validated in a natural history study in children and adolescents with DM1 and AMO plans using it as the primary outcome measure in its upcoming registration trial. [!\[\]\(c50c8b7b2cc2cf9ff925edec0ee94c0d\_img.jpg\)](#)

## Losmapimod to be trialled in FSHD

Targeting the root cause.

Fulcrum Therapeutics, a biotechnology company focused on discovering and developing therapies to rebalance gene expression, has partnered with GlaxoSmithKline (GSK) for the development and commercialisation of the investigational drug losmapimod. There are currently no approved treatments for facioscapulohumeral muscular dystrophy.

“Losmapimod is a foundational clinical asset for Fulcrum that has the potential to become the first approved therapy that targets the root cause of FSHD. Fulcrum believes losmapimod has the potential to slow, or halt, the

progressive muscle weakness that characterises the condition, which would significantly improve patients’ quality of life,” said Robert J. Gould, Ph.D., Fulcrum’s president and chief executive officer.

“The agreement shows confidence in our unique approach to rebalancing gene expression in severe genetically defined disorders. We will work urgently to advance the compound through the clinic.”

Fulcrum identified inhibitors of mitogen activated protein kinase (MAPK) as powerful inhibitors of DUX4 expression. DUX4 is the gene that is the root cause of FSHD. Losmapimod is a selective MAPK inhibitor that GSK has tested extensively in clinical trials, but never in muscular dystrophies. Fulcrum’s novel insight into the DUX4 regulatory pathway led the team to review existing MAPK inhibitors, and Fulcrum identified losmapimod as a compound with the potential to address the root cause of FSHD by decreasing DUX4 expression.

GSK evaluated losmapimod in more than 3,500 healthy volunteers and patients in 24 clinical trials across multiple indications. The data has provided evidence that losmapimod is a well-tolerated agent. Fulcrum has conducted preclinical testing of losmapimod in patient-derived cell models and observed precise and potent downregulation of DUX4 expression and restoration of a healthy muscle phenotype without an effect on myogenesis.

Fulcrum expects to initiate a Phase 2b clinical trial of losmapimod in patients with FSHD at multiple clinical sites in the U.S. and Europe in mid-2019. [!\[\]\(166772600a13ad0a433053f90fe45649\_img.jpg\)](#)

# NF 1 – Neurofibromatosis 1

Research into improving treatments and finding cures for NF1 is ongoing but as neurofibromatosis cannot currently be cured, the goal is to monitor the condition and intervene when treatment is required.

Neurofibromatosis is a genetic condition that affects the skin, soft tissue, bone and nervous system. It is characterised by the development of soft tumours called neurofibromas that develop on the nerves and brain, or grow on, or under, the skin.

As they grow, the tumours can press on important areas in the body, affecting the way the body functions with varying degrees of severity. Neurofibromas are usually non-cancerous but in a small percentage of cases may become cancerous.

Neurofibromatosis is a genetic condition and can be passed from a parent to their child. However, about half of all cases are due to a new mutation in the gene that causes NF1.

NF1 is the most common type, representing about 90 percent of all neurofibromatosis cases. In New Zealand it is estimated that NF 1 affects about 1 in 3,000 people. It is caused by a defect in a gene on chromosome 17.

NF 1 may also be called von Recklinghausen disease – named for a German pathologist, Friedrich Daniel von Recklinghausen who, in 1882, first characterised the tumours in neurofibromas which consist of mingling of nerve cells and fibrous tissue.

NF1 is characterised by:

- Café-au-lait spots (light brown patches on the skin).
- Multiple neurofibromas.
- Freckling on the skin under the armpits and in skin folds eg: the groin.
- Tiny tumours in the iris of the eye (Lisch nodules).

NF 1 is usually diagnosed in infancy or early childhood.

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***Cutaneous neurofibromas are superficial, soft tumours that grow on the surface of the skin.***

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## Signs and symptoms

The severity of the condition tends to vary greatly from person to person. Some people are barely affected by the condition, while others are severely disabled by it.

Approximately 60 percent of people with NF 1 will have only relatively mild signs and symptoms.

The most obvious signs of NF 1 are those that affect the skin – café-au-lait spots, freckling, and neurofibromas of the skin. Café-au-lait spots usually measure at least 5mm in diameter in children and can grow to 15mm in diameter in adults.

Neurofibromas are the hallmark of NF 1. There are four main types of neurofibromas, each classified by where they develop in the body.

Cutaneous neurofibromas are superficial, soft tumours that grow on the surface of the skin. Subcutaneous neurofibromas grow within the skin layers and can cause local tenderness. The other two types, called nodular plexiform and diffuse plexiform, develop within the body and can cause problems such as:

- Curvature of the spine (scoliosis).
- Malformation of the long bones below the knee and elbow.
- Vision loss.
- Bleeding or blockage in the gut.
- Seizures.
- Hydrocephalus (accumulation of fluid on the brain).
- High blood pressure.

Children with NF 1 have high rates of speech impairment, learning difficulties and attention deficit hyperactivity disorder. Puberty may be



early or delayed. Some cancers occur more commonly in people with NF 1. These include some malignant brain tumours, leukaemia, and cancers of the muscles, kidneys and adrenal glands.

## Diagnosis

A diagnosis of neurofibromatosis is based on the characteristic signs of the condition and certain diagnostic criteria. A careful personal and medical history will be taken. For a diagnosis of NF 1 to be made, two or more of the following signs need to be present:

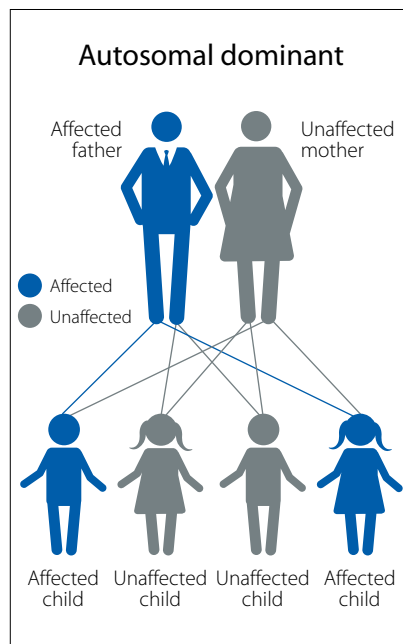
- Six or more café-au-lait spots larger than 5mm in diameter
- Two or more neurofibromas
- Freckling under the arm or around the groin
- Lisch nodules
- A tumour on the optic nerve
- Certain abnormalities of the skeleton
- A family member with NF 1.

## Treatment

Research into improving treatments and finding cures for NF1 is ongoing but as neurofibromatosis cannot currently be cured, the goal is to monitor the condition and intervene when treatment is required.

As a general rule, tumours that do not cause any problems are usually left alone. If treatment is required, it usually involves surgical removal of tumours. Surgical removal may be performed if:

- Tumours cause discomfort and pain.
- Tumours keep getting irritated or damaged.



Mode of inheritance for neurofibromatosis type 1.

- Tumours in the spine or brain are causing symptoms.
- Sensation or motor function is affected.
- A tumour is suspected, or known, to be cancerous.
- Skeletal abnormalities, such as scoliosis, may also require surgical treatment.

If tumours are disfiguring, plastic surgery can help to improve cosmetic looks.

Physiotherapy can be useful if mobility or function is affected. If there are deformities of the feet and/or legs orthopaedic devices can also help to improve mobility.

Some brain tumours or tumours along the nerves can be treated with chemotherapy or radiotherapy.

People with neurofibromatosis should have regular physical and neurological examinations. Attention

should be given to monitoring changes in the size or number of neurofibromas.

Referral to Genetic Health Services is also useful.


## Genetics of NF1

Approximately half of individuals with NF1 inherit the condition from one of their parents. The other half develop it as the result of a spontaneous (also called new or de novo) change in one of the NF genes in the egg cell or sperm cell during conception.

NF1 is inherited in an autosomal dominant way. This means that individuals with NF1 have a 50 percent chance of passing the condition on to their offspring. If a child inherits the NF gene, they will always show symptoms of the condition, however, it is difficult to predict the severity. This is called variable expressivity.

If a child of someone with NF does not inherit the NF gene, they do not have NF and therefore cannot pass it on to their future children. That is to say, NF cannot "skip a generation".

## Research

There are several drugs that are being trialled looking to treat various aspects of NF1 and NF2. Visit [clinicaltrials.gov](https://clinicaltrials.gov) and enter NF1 into the keyword search. 

## Just getting on with it

Twenty-year-old Amber Trembath and her family are very proactive in dealing with Amber's diagnosis and Amber is currently on the look-out for part-time, flexible work. Here Amber, and her mother Susan, share Amber's journey with Neurofibromatosis Type 1.

I was checked thoroughly for any defects when I was born, as my mother had done with her first daughter Hannah, because my dad had an extra finger when he was born. The only thing my parents could find with me, was a large coffee-coloured patch of skin on my pelvis. My parents asked the doctors what it meant and they said 'not to worry, it's nothing'.

When I was eight weeks old, my dad went to a skin specialist as he had bumps over his body and wanted them checked out, not knowing what they were. The skin specialist said he had Neurofibromatosis and asked if any of his children had coffee coloured patches. My parents said Amber has a big one and the skin specialist said, "watch her."

(Note: Coffee coloured patches, also known as café au lait spots, or café au lait macules, are flat, pigmented birthmarks. The name café au lait is French for "coffee with milk" and refers to their light-brown colour. Café au lait spots are often harmless but may be associated with syndromes such as neurofibromatosis type 1 and other conditions.)

### About Neurofibromatosis

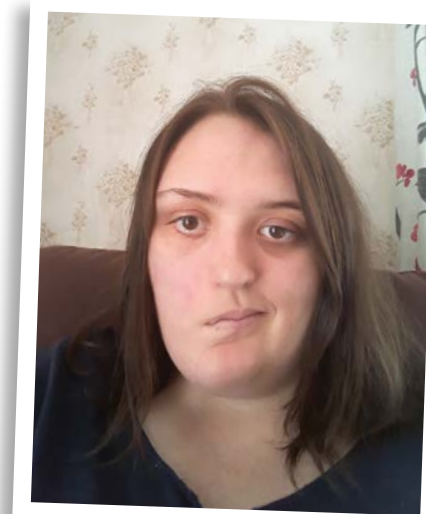
Neurofibromatosis type 1 is the most common type of neurofibromatosis.

It affects one in every 3,000 people born worldwide. The cause of NF1 is a change in a gene. A gene is a sequence of DNA, which is the genetic code that is passed down from parents to children.

***My parents asked family if anyone else had NF, but my dad is the first one, so he is a spontaneous mutation, which has then been passed on to me.***

Because of this, a child can inherit NF1 from a parent who has the disorder. About half of the time, however, a child with NF1 is the only person in the family who has the disorder. In such instances, the NF1 gene change occurred as the result of "spontaneous mutation"—a random error in the process of copying genetic information.

Neurofibromatosis 1 is not the consequence of drug, alcohol, or X-ray



Amber Trembath

exposure, or any other factor under the control of the child's parents. NF1 is not contagious. Contact between an affected child and an unaffected child cannot transmit the condition.

The NF1 gene is responsible for the production of neurofibromin, a protein that keeps cells from growing too quickly. Without this control, the cells can continue to grow and divide around the nerves, producing the tumors commonly found in NF1.

Neurofibromin is also involved in processing nerve signals, including signals inside the brain and those that send messages between the brain and the muscles.

That is why problems with neurofibromin affect how the brain receives, processes, stores, and sends

information. It is difficult to predict the progression of symptoms, so it is recommended that people see their doctor regularly. There is still much left to learn about NF1. The medical community is working diligently to gain a full understanding of this condition.

(Sourced from NF Parent Guidebook CTF web 2017.pdf)

After digesting similar information to this from the skin specialist, a friend of the family copied a whole NF website so my parents could read some more about it.

My parents asked family if anyone else had NF, but my dad is the first one, so he is a spontaneous mutation, which has then been passed on to me. My sister Hannah is not affected at all.

I had issues with chronic ear infections, so my parents were taking me to the doctors, a lot. An ENT at the hospital said there was something white behind my right eardrum and they would like to operate and see whether it was bone or something else. My mother asked if it could be NF. At 18 months old I had surgery to lift the eardrum and the report that came back said, mother is right: It's NF. Then the tumour spread to what it is today through both sides of my neck, right side of face, skull and spreading to my left side.

I have had several more operations to debulk my neck, plastic surgery to debulk my face and lift up my right ear (which keeps dropping). The same tumour has also eaten away bone in my skull, so I have a mesh plate to protect my brain, where the hole was.

I also have several other tumours, one near my spine, a few in both legs but so far they have been behaving. More recently I have a small tumour on my tongue.

## School days

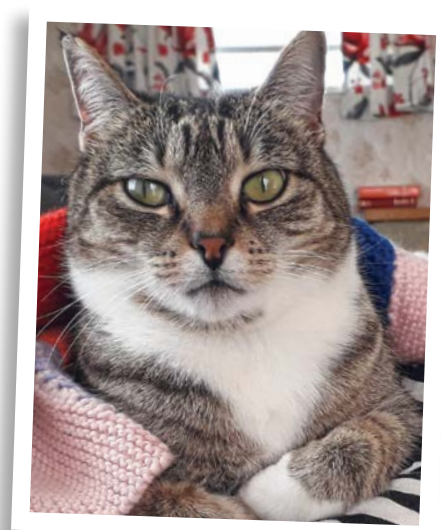
It was very difficult for me at school, I was often sick, or in surgery or seeing doctors/surgeons. The children at primary school thought I was just having fun, bunking off school.

A child bullied me and my parents got straight onto it with the staff and the bullying stopped. And my mother was invited to talk to the class about my medical condition, which went really well and those children then fielded questions from other children in the playground.

I missed camps, swimming and belonging to team sports. My mother used to give the teachers a list of all of my upcoming appointments, so that they had an idea of when I would be absent. Wellington Hospital has the Central Regional Health School where the teacher co-ordinates school work when a child is sick in hospital and that was fantastic. It meant I could keep up, to a certain extent, with my peers.

After college I went on to Weltec in Petone to do a couple of food courses which I passed. Currently I am looking for part-time flexible work, which I can fit my appointments around.

In the meantime, I like playing with my (therapy) cat Sophie, playing word games on my laptop, walks and knitting.



Sophie, Amber's pet cat.

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***One doctor said she was stoic, I reckon that is a great word to describe Amber. She just gets on with it.***

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## Amber's mother, Susan

It was so hard watching Amber go through surgery and being in pain. Over the years I've learnt so much about NF and Amber's body and she has a very high pain threshold.

One doctor said she was stoic, I reckon that is a great word to describe Amber. She just gets on with it. We've had a few meltdowns over the years but seem to get through it.

We haven't been successful in finding a pain medication that suits her and that doesn't have bad side effects (like being drowsy or spaced out). Only now, when she is 20, is she





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**Post:** Make a donation by post. Our postal address is: PO Box 12063, Penrose, Auckland, 1642

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*Thank you. We greatly appreciate your support.*

actually in communication with the pain clinic, and waiting for her first appointment.

Amber's speech has been a big issue, because of Bell's Palsy that she got post-surgery. Unfortunately, in New Zealand, there is a great shortage of speech language therapists (SLT's) so Amber only had one for a short time. This service stopped for her at aged eight.

## Useful tips

- Dealing with doctors: Well they don't teach you how to do that at school, so that has been a learning curve. At first it was a bit intimidating and I felt I didn't know enough to be a good advocate for Amber, but after explaining to the doctor "could you please simplify the medical jargon, so we can understand it", discussions have since gone a lot smoother.
- Be prepared, have a list of questions to ask.
- If you need more time with the doctor ask for a double appointment.
- Keep your own notes on your child. Amber had a tickle cough for a year and the doctor tried asthma inhalers, antibiotics, steam and it turned out to be two cancerous tumours behind her throat. So she had to undergo surgery and radiation, that was five years ago. Thanks to my note taking I could discuss it better with the doctor and get further investigation done.
- Also keep a list of operations - date, what procedure, why, where. Also scans and other tests and anaesthetics (this list should roughly match the surgery list), and any implanted objects.
- I've found over the years with scans and operations and the forms you have to fill out, that I just put "see list", and make sure that I have a copy of these lists to give them. It also saves a lot of time.
- After an anaesthetic - don't give fizzy drink to the patient, unless you like big messes. Water is best, little sips.
- Don't leave the hospital until the patient can safely walk.
- If you are supporting someone having an anaesthetic or scans - take a book or knitting with you. One surgery we took a scrabble game to play while we waited.

---

***Dealing with doctors:  
Well they don't teach  
you how to do that  
at school, so that has  
been a learning curve.***

---

## Amber and MDANZ

MDANZ has been a good support group to belong to. The field officer, Dympna was good to talk to, the MDA *In Touch* magazine is full of useful information and other people going through difficult journeys too.

# Personal goal setting

## *Planning to live your life, your way*

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The time you spend figuring out your abilities and the options for reaching your personal goals is a great investment in improving your situation and moving towards the life you want.

What comes to mind when you think about the next step in your life? If you agree that you want to build a more stable future, then you have to include looking and preparing for opportunity.

Despite challenges you face and obstacles that may undermine your current situation, there are plenty of things you can explore and research to try to create opportunities for success.

And the time you spend figuring out your abilities and options for making progress towards your goals is a great investment toward improving your situation and moving towards the life you want.

The first step is to have a thorough understanding of where you are today and where you want to be.

You can begin by reviewing aspects of your life today and then identifying a goal that you would like to reach in that area.

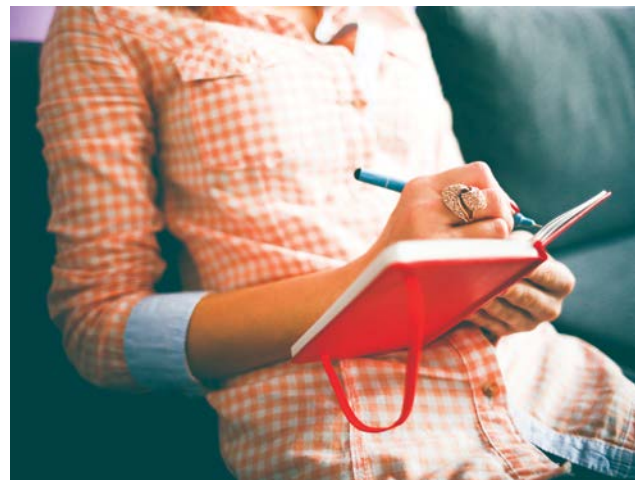
Some people like to do this type of personal review with a family member or friend. And that can be helpful in providing a reality check and in coming up with additional ideas you might not think of on your own.

If you prefer to do this by yourself, that's okay, too.

### How to set a goal

- First consider what you want to achieve, and then commit to it.
- Set goals that motivate you and write them down to make them feel real.
- Then plan the steps you must take to realise your goal.
- Cross off each one as you work through them.

Once you've decided on your first set of goals, keep the process going by reviewing and updating your to-do list on a daily basis.



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*Once you've decided on your first set of goals, keep the process going by reviewing and updating your to-do list on a daily basis.*

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Periodically review the longer-term plans and modify them to reflect your changing priorities and experience. (A good way of doing this is to schedule regular, repeating reviews using a computer-based diary.)

When you've achieved a goal, take the time to enjoy the satisfaction of having done so.

Absorb the implications of the goal achievement and observe the progress that you've made towards other goals.

If the goal was a significant one, reward yourself appropriately.

All of this helps you build the self-confidence you deserve.



## How can your fieldworker help you?

DYMPNA MULROY

Friendship, encouragement and support are all part of the fieldworker service. Here are some stories from members on their experiences with our fieldworkers.

The theme for this *InTouch* issue is 'goal setting'. This has been reflected in our member stories with the great achievements they have accomplished.

Goals can be short or long term, such as baking a cake or going on an overseas adventure.

The resources, support and funds required will depend on the goal you set.

Discussing your goals and ambitions with others affirms your decisions and makes it real.

It can be helpful to discuss these goals with your local fieldworker who can support you in achieving them.

The fieldworker service is nationwide. It doesn't matter where in the country you live the fieldworkers can travel to meet up in your community.

Here are some stories from members who have used the fieldworker service in the past.

If you would like to get in touch with your local fieldworker you can locate the branch contact details on the inside back cover of this magazine.

"I made contact with the fieldworker service for general support six years ago.

"Since then I have engaged in the service numerous times for support and advice on different things.

"This includes applications to the Community Lotteries for a mobility vehicle, an application for a medic alert bracelet and support letters in an advocate role.

"My fieldworker has supported me when relocating to accessible homes and worked with my

***I always felt listened to and supported. It was good to have someone I could confide in and know my privacy would be respected.***

occupational therapist when my house was being modified.

"I always felt listened to and supported. It was good to have someone I could confide in and know my privacy would be respected.

"I believe the fieldworker service is an essential service in MDANZ. It is important to have this support available to members.

"I will continue to access this support as needed throughout my journey with a neuromuscular condition." – Wayne.

"I found the fieldworker service to be extremely helpful and supportive.

"When my fieldworker got in touch, it was wonderful to have support from someone who took the time to care and listen.

"It was helpful to discuss my situation with someone who had knowledge and experience as I was transitioning through some difficult changes at the time. She understood the difficulties and was a great encouragement.

"I accessed the fieldworker service because I needed support. Having been discharged from the hospital system and from ACC rehabilitation, I was on my own. Without someone who knows how the system works to guide me, it felt like a maze at the time." – Eric.

"My wife Janine has Myotonic Dystrophy and Dympna was our Wellington MDA regional fieldworker from May 2012 through till Sept 2018.

"Janine and I both highly enjoyed and valued her friendship and



support during that time and, since her departure, we have missed being able to call her and talk about matters associated with Janine's condition.

"Our fieldworker has been so helpful to us over the years. A summary of the help and support provided by our fieldworker includes:

- Regular email updates to keep us informed on local and national events, encouraging networking and friendship with others who have similar conditions

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***Regular email updates to keep us informed on local and national events, encouraging networking and friendship with others who have similar conditions and updates on national and international research and developments.***

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and updates on national and international research and developments.

- Supporting Janine and I with visits to see how things are going and if additional assistance/support is needed, and to hear and give encouragement.
- Referrals to hospital occupational therapy for support rails etc to be installed in the shower and by the front door steps.
- Arrange for CAM (cough assist machine) loans and getting the appropriate training and sign off



by hospital occupational therapy/physio staff each time.

- Advice on sleep apnea and therapy and who to contact for assistance.
- Encouragement and follow-up support to Janine in using her VPAP (variable pressure breathing machine).
- Advice on pneumonia vaccine for Janine funded by MDA.
- Hospital visits when Janine was sick in hospital which greatly boosted her recovery."

– Martin and Janine.



*Dymrna Mulroy is the new member services manager at MDANZ.*



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## 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 1950s. 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 core team



*Trevor Jenkin*  
National Executive  
Chairperson



*Miriam Rodrigues*  
Programme and  
Service Advisor



*Brian Hadley*  
Accountant and  
Business Manager



*Dymrna Mulroy*  
Member Services  
Manager



*Natalie Foote*  
Marketing and  
Communications Manager

### Northern Branch



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

### Central Branch

Fieldworkers  
Ph: 0800 886 626  
Email: info@mda.org.nz

### Canterbury Branch



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

### Southern Branch



Fieldworker: *Jo Smith*  
Office Manager: *Vivienne Fitzgerald*  
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

Michael Schneider. Ph: 021 851 747  
Email: spider@spider.co.nz

### Central Branch

Bernadette Ingham. Ph: 027 600 3868  
Email: members.central@mda.nz

### Southern Branch

Matthew Willetts.  
Email: willetts.matthew@yahoo.com

### Canterbury Branch

Jan Daly. Ph: 027 686 1152  
Email: mickandjan@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  
Schwannomatosis  
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



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