InTouch

KIA NOHO TATA Inclusive | Inspiring | Informative Autumn 2017 Issue 94

The power of support
We are stronger together

Finding hope
A diagnosis changed my life

Chasing adventure
An exciting programme for young people

A day in a fieldworker's life
Working with members, families and whānau

Muscular Dystrophy New Zealand

InTouch

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Catching up with Heather

Kia ora koutou, greetings to you all.

What a pleasing and exciting year 2016 was for the Muscular Dystrophy Association. I am very proud to have been able to work with a very supportive National Council, and alongside strong Branch networks to support Ronelle, and her team to deliver such great outcomes.

Ronelle and Brian have also done a sterling job of managing our business and financial commitments. They have ensured that administrative costs have been tightly managed and, wherever possible, reduced. This means that our precious resources can be channelled into services for members. I am sure, like me, they have enjoyed the guidance and insights offered by Ken Green, National Council member and Chairperson of the Finance Committee. Ken's clear vision and financial skills have proven to be a winning formula in this role and it has been a real delight to work with him.

So not only did we end the year with a pleasing financial surplus (which can be invested back into member services), we were able to support 38 applications to the members' discretionary fund. The fund of $21,500 has been distributed to eligible applications across three funding rounds this year. This is another very tangible demonstration of the Muscular Dystrophy Association's support for members.

The other significant achievements last year included the revitalising and update of our Strategic Plan, the associated rebranding, and the very successful Freedom Campaign. We can collectively feel really proud of the participation and feedback from members in all these activities. It has ensured we are presenting a consistent, inclusive, vibrant, and professional image of the Muscular Dystrophy Association in all forums in which the Branches and the National Office are involved. This is important, not only to uphold our position of respect in the sector, but also to attract members, and to draw the attention of financial supporters and sponsors.

To conclude, I remind you that this is an important time in the Muscular Dystrophy Association calendar. With the various AGMs being held in between now and April, it is time to consider whether you would like to be involved in the governance and strategic leadership of the Muscular Dystrophy Association. Think about it – everyone has skills and expertise to bring to the table and this is an opportunity to shine your leadership light on your own organisation.

Warmest wishes to you all

Heather Browning
MDA Chairperson

In touch with Ronelle

Tēnā koutou katoa, greetings to you all.

Google leadership quotes and you'll get millions of thought provoking options. One that I really relate to is simple and doesn't need a lot of external resource to achieve; "*The most powerful leadership tool you have, is your own personal example.*" It's by the late John Wooden, a dedicated US pro basketball coach.

I've been reflecting on this quote as we settle into the first quarter of 2017, and planning gets underway for the annual general meetings for each of the branches and for the national body.

These forums present an opportunity to report on the outcomes achieved for the 2016 financial year, and to identify how our new core values can strengthen the organisation for the year ahead. It is also a time that we call for new leaders.

Last year I asked you, my fellow colleagues and members, to consider the important role of governance for our organisation. Many of you stood for election, voted and contributed to the process. The voter turnout in 2016 was slightly higher (12%) than it was for 2015 (10%) and we have had a great year and a strong National Council leading and working alongside the MDANZ team. Thank you.

This year, I want to encourage you again to participate in the process, which takes a lot of money and time to administer. You may want to nominate someone with skills and passion for our organisation and what it stands for, vote for the amazing candidates who agree to be nominated, or consider standing yourself for National Council or your Branch Committee.

This association has been supporting Kiwi families like yours and mine for almost 60 years, and with your help, we can ensure a sustainable future for this organisation, so it can be here for those yet to come. MDANZ has a vital role to play in advancing policy, research and access to treatments, as well as providing individuals and whānau with information, support and resources. We can make an even bigger difference if we lead by example.

Ngā mihi mahana,
Warm regards

Ronelle

Ronelle Baker
Chief Executive

Richard Roxburgh and I are doing Auckland's Round the Bays on Sunday March 5th to raise money for research. We'd love you to join us.

A cup of tea and a catch up with … Marty Price

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 been with MDANZ since September 2014 as fieldworker for the Nelson, Marlborough and West Coast regions.

What qualifies as a great day at work for you?

Knowing that I contributed my all when supporting our members and that the necessary tasks were done to enable our members to live a healthier and more independent life.

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

Having an equal opportunity to access any equipment they may require. I'd also like to see a community where access is not a barrier, but gives freedom to live without the worries and stress many live with on a regular basis.

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

Sausage rolls and coffee.

What are you passionate about?

My beautiful children – seeing them grow and develop. I also like to make sure my friends get together and have fun. Another passion is volunteering in the community. I am in my 10th year with Community Patrol of New Zealand and really enjoy assisting the Police and the community.

Save the date

APRIL 28

The Annual General Meeting of MDANZ will be held on Friday 28th April from 6-9 pm at Distinction Hamilton Hotel and Conference Centre, 100 Garnett Avenue, Te Rapa, Hamilton.

Join us for an evening of engaging speakers, and mix and mingle with your governance, staff and other MDANZ members.

Free parking is available and refreshments will be provided.

Branch AGMs

Northern: Saturday 8th April, 3pm at the MDANZ National Office, 419 Church Street, Penrose, Auckland. Speaker and afternoon tea provided.

Wellington: Sunday 26th March 2-4pm at the Walter Nash Stadium in Taita, Lower Hutt.

Christchurch: Saturday 18th March, 1pm, in the boardroom at the Papanui Library.

Southern: Saturday 4th March, 1pm in the MDANZ Office 8 Baker St, Caversham, Dunedin.

Contact your branch if you need help with transport costs. We look forward to seeing you.

Funds for freedom

It has been a great first year of the Members Discretionary Fund. This was launched in April 2016 for our members to have access to funds for resources that would support them to experience more freedom in life.

Seventy members applied throughout the year and we were able to make a contribution to 35 people. They were of all ages and from all over the country. Contributions have been given towards education, gym, mobility equipment and scooters, travel and power generators.

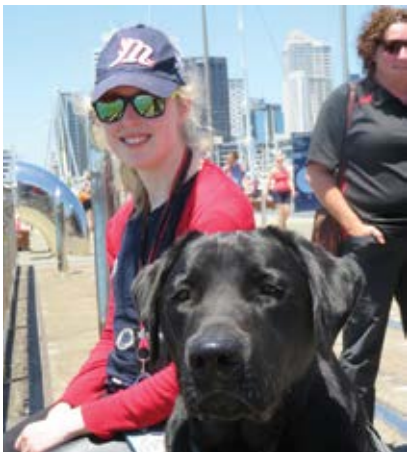
Trevor Wilson, from Motueka, has contributed to Canterbury Branch camps for several years, singing and performing with his guitar. He was seeking funds towards the purchase of a detachable mobility scooter that he could take in his motor home when he travels, allowing him to fully experience the trips by being able to get out and about. It took Trevor two goes to get funding, and he didn't receive all that he asked for, but Trevor is grateful for the contribution towards his passion for travel.

Trevor Wilson

Benjamin Ragland has a passion for woodwork and produces trestle tables. After his tools were stolen in a burglary, he had to use an old saw that made his job very labour intensive. With the help from the discretionary fund, he was able to buy a piece of equipment that would make it easier for him to perform his handiwork and manage the workload better.

Therese Boon and Damian Duffy both applied to the fund for the same goal – a contribution towards the placement costs of a Mobility Dog. They had both successfully progressed with Mobility Assistance Dogs Trust who trained dogs ready for placement with just the right people. Therese was thrilled to be matched with Newton, a black Labrador and knows they will grow together to meet the changing needs of her progressive condition.

Damian is equally thrilled with his dog Hunter, and wrote to thank MDANZ for the contribution. "As life goes on new challenges will arise and with muscular dystrophy sometimes it's hard to understand yourself, let alone trying to explain it to somebody else. In Hunter I have a companion to help me through the tough times and help me adjust to whatever my circumstance might be in the future," he wrote.



Above: Therese with her dog Newton and Damian with Hunter.

New member Aden and his mum Tracey were grateful for the contribution they received towards the cost of attending a Scout Jamboree in Blenheim.

"The jamboree was an action packed 10 days ... and I made many memories I will never forget".

For more about the fund including a list of recipients, go to www.mda.org.nz

Webpage for Paediatric Neurology Network

A new resource to help deliver better care for children

The Paediatric Neurology Clinical Network now has its own webpage on the Starship website! www.starship.org.nz

The purpose of the Paediatric Neurology Clinical Network is to support multidisciplinary teams who work across NGO, primary, secondary and tertiary services, to deliver best practice care and treatment of children with neurological conditions throughout New Zealand.

Children's Neurology or Paediatric Neurology refers to a specialised branch of medicine that deals with the diagnosis and management of neurological conditions in children from birth to adolescence. The discipline of Paediatric Neurology encompasses childhood diseases and disorders of the brain, spinal cord, peripheral nervous system and autonomic nervous system.

The care of children with neurological conditions is delivered in a wide range of settings. With tertiary paediatric neurology centres in Christchurch, Wellington and Auckland this Network aims to enhance co-ordinated management across the entire continuum of care. A key part of the workforce is education and development.

Currently the network is supporting the development of the following guidelines;

- NZ Paediatric Epilepsy Guidelines
- Allied Health & Nursing Best Practice Guidelines for Duchenne Muscular Dystrophy

And is in the process of endorsing a number of guidelines around the care and management of Duchenne muscular dystrophy.

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**

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

Bequests: You can create a lasting difference through making a bequest. Contact us or visit our website for information on how to include MDANZ as part of your will.

Thank you. We greatly appreciate your support.

Granted!

The 2016 funding round of the Neuromuscular Research New Zealand received eight applications with three projects being granted all or partial funding.

Some of the applications that unfortunately missed out on receiving funds were considered by the trustees to be 'near miss' applications and it was only due to the limited nature of the funds available that meant that this year the trust was unable to support them.

The projects that received funding are:

- The New Zealand Neuromuscular Disease Registry led by Dr Richard Roxburgh and co-ordinated by Miriam Rodrigues.
- Respiratory Care Pathways led by Alister Neill, Associate Professor of Respiratory and Sleep Medicine and Director of WellSleep for the University of Otago's Department of Medicine in Wellington in collaboration with Dr. Tristram Ingham Senior Research Fellow at the University of Otago, Wellington.
- A postgraduate scholarship was awarded to Shelby Taylor, Master of Science student in Genetic Counselling at the University of Melbourne for a project investigating family communication in myotonic dystrophy.

More about these projects in future issues of *In Touch*.

We acknowledge funding from Richdale Charitable Trust for making these grants possible.

Miriam Rodrigues and Dr Richard Roxburgh.

Dr. Tristram Ingham

Shelby Taylor.

A quick questionnaire

The NZ NMD Registry has a new way for people with myotonic dystrophy to become informed about the Registry, consent to participate, update details and provide relevant clinical information about yourself.

Simply enter the link below into a web browser, and scroll!

https://nmdregistry.org.nz/rdrf/DM1/questionnaire/nz

The questionnaire will probably take you between five and ten minutes to complete. We really value your input and hope that by collecting important information about you we will be able to bring more research and clinical trials in myotonic dystrophy to New Zealand so tell your family about it too!

Although the questionnaire is currently called "DM1" it is for all types of myotonic dystrophy, including congenital myotonic dystrophy, classical myotonic dystrophy (DM1) and myotonic dystrophy type 2 or DM2 also known as proximal myotonic myopathy or PROMM.

If you have any questions please contact your MDANZ fieldworker or the Registry Curator registry@mda.org.nz

BRANCH news

Catching up with news from around the country

Canterbury

We are excited about our upcoming camp planned for 2nd – 5th March at our favourite spot in Hanmer Springs. This year's camp will be for families including children, as our camp for young people had to be cancelled last year.

Our Office Manager, Eris Le Compte, has announced her retirement and will be leaving the Branch following more than a decade of service. We wish Eris all the very best for the future and look forward to introducing our new Office Manager in the next issue of *In Touch*.

Members of the Canterbury Branch enjoying the end of year BBQ held at The Groynes in December 2016. Those who attended had a great time with superb food and lovely weather.

Farewell Sue

It was with a heavy heart that we said goodbye to one of the pillars of the Canterbury Branch. Sue Robinson passed away suddenly over the Christmas period, and we are saddened by this loss.

Sue will be remembered for her tireless giving, generosity and commitment to MDANZ. She was a devoted parent to her son Blair who had DMD. Through Blair she became involved with MDANZ, held the position of Chairperson for the Canterbury Branch for many years, and served on the National Council.

Right up until her death she was a major fundraiser and organiser on our local committee, raising thousands of dollars for the Canterbury Branch. Sue was a Canterbury finalist two years ago for the New Zealander of the Year Award, and was a life member of MDANZ. We express our sincere condolences to her husband, Ken, and family.

mdacanty@xtra.co.nz

Sue, in 2009 as a member of National Council.

Wellington

Are you prepared?

All of New Zealand is at risk of earthquakes, but in the Wellington region we've been particularly reminded of our vulnerability recently. We can't predict when one will happen, but we can take steps to protect ourselves and our family.

In the event of an earthquake or other natural disaster, it is important for those who have, or have a family member with, a disability to ensure an appropriate plan is in place.

There are resources online to help. Civil Defence advises the creation of a personal support network with a minimum of three people to alert a person with a disability of civil defence warnings, or to help if the need to be evacuated arises. Civil Defence recommends those who are unable to take cover under a table, move near an inside wall away from windows and items that could fall, and if in a wheelchair to lock the wheels' stopping movement. More information, including the Drop, Cover and Hold Information Sheet for people with disabilities, is available at getthru.govt.nz

This webpage also has useful tips on things to put in an emergency kit and what needs to be considered when you or a loved one has a disability. getthru.govt.nz/how-to-get-ready/people-with-disabilities/

Emergency Management Canterbury published a resource *Disaster Preparedness for People with Disabilities*. It was designed to help people who have physical, visual, auditory or cognitive disabilities to prepare for natural disasters and their consequences. Although it was published in 2009, the information is still very helpful. http://cdemcanterbury.govt.nz/media/9969/disaster_preparedness_book.pdf

Wellington Branch has First Aid kits available at the office. The cost is $45 each/$50 including delivery. Each Kit contains 77 items including a digital thermometer and two survival blankets. We receive $12 from every kit sold.

Contact the Wellington office on 0800 886626 or office.mdawgtn@xtra.co.nz if you would like to order one.

office.mdawgtn@xtra.co.nz

Southern

We have lots planned for our members this year. Our Fieldworker Jo Smith has been accredited to run change, grief and loss workshops named Seasons for Growth. The Seasons for Growth programmes were specifically developed to address the needs of adults and children to understand and manage change, loss and grief. We plan to pilot this programme in Otago and Southland. If this is something that you would be interested in contact Jo on 027 509 8775 or email joanne@mda.org.nz. All enquiries are confidential.

Providing social connection is certainly one of the items on the top of our list. We would like to set up support groups in all areas. If you would be interested or have ideas of potential meeting places in your area we would certainly be interested in hearing from you. We have great facilities here at our offices at 8 Baker Street, Dunedin that we can use.

We also have a Facebook page www.facebook.com/MDASouthernBranch Please 'like' and 'share' our page. We will be posting events throughout the year.

Brain awareness week starts on the 12th March and runs for the week. We will be participating and are very fortunate to be having a guest speaker from the Brain Health Research Centre presenting on the 17th March at our rooms. Save the date!

joanne@mda.org.nz

Northern

We had the second of our two camps in November, with around 50 people attending, including lots of new families. It was also a good opportunity to combine our Christmas party with the camp and around 100 members attended, which is a record. Everyone had a great time.

support@mdn.org.nz

Fieldworker Darian Smith catches up with Naera Haronga over coffee

A day in the life of a fieldworker

Here for members, families and whānau

Our fieldworkers work in the community providing personalised support and education for members with lived experience of a neuromuscular condition. Find out what they may do on any given day.

Sometimes it's easy to forget that a fieldworker isn't just there to work with the person who has the condition, but those around them as well. This can be family, and it can also be medical professionals, employers, schools and peers. Darian Smith, Northern Branch, talks about school visiting and attending clinics with members.

School visit

I often visit schools where a young member attends as a student. Part of the support we offer is to provide educational sessions about the impact of neuromuscular conditions for children and young people. These talks can be either for the school staff, or for the students, to raise awareness and support student participation in school life.

When talking to staff, I will sometimes use a PowerPoint presentation or, if it's a small group, simply provide them

with information and discuss with them what some of the experiences can be like living with a progressive neuromuscular condition, and how they can make their student's time at school easier. Usually parents have already discussed the situation, but school staff can sometimes find it difficult to understand the nature of neuromuscular conditions – especially the delayed impacts of physical activity such as fatigue and how that can be for the student in a school environment. School staff seem to value the opportunity to talk to someone neutral and ask questions.

When I'm planning to talk to the students themselves, I first ask the child with the condition how they would like to be involved. Some children like to be in front of the class presenting with me, others are happy to be named and discussed but some prefer not to be singled out or even not to be present at all. Any of these options is totally fine.

The session usually begins with a discussion about 'difference'. I point out that some of us need glasses to help our eyes see better, and neuromuscular conditions are like that but for muscles. I give an age appropriate outline of the condition, talk a bit about genetics and how conditions like this don't transmit to other people unless there is a family connection. I like to use a fun physical activity to give the children an experience of what it might be like to have muscles that don't work as well as they're used to. I ask them to brainstorm ways they could be inclusive and caring of someone with a neuromuscular condition and answer any questions they may have.

Studies suggest that peers who are educated about a student's condition are more supportive and less inclined to bullying and the anecdotal evidence from our members also supports this. It's a great way for us to make a tangible improvement to the school experience for someone with a neuromuscular condition.

Clinics

Some of our members attend neurology clinics. One of the ways I support members and medical and allied health professionals is to attend these clinics as well. Clinics vary across locations and may have a different focus. Some will be a simple appointment with a neurologist. Others will include a multidisciplinary team that may include a physiotherapist, occupational therapist and speech language therapist.

Studies suggest that peers who are educated about a student's condition are more supportive and less inclined to bullying ...

Having a fieldworker at clinic provides a variety of benefits. I'm there as a friendly face and support for the member, I can help prompt information and issues if necessary, and can be a good source of information for any health professionals who are new to the neuromuscular field. Even more importantly, the ability to be adaptive and holistic in approach means I can follow up on a variety of issues that come up, which the doctor may not have the time or ability to deal with. For example, support and information on funding sources, social supports, school support, green prescription, employment, Total Mobility card and many other issues may come up in the course of a clinic appointment but be outside the purview of the health professional to do much about within the allotted timeframe of the appointment. As a fieldworker, I can make suggestions and provide ongoing support outside of the clinic, and ensure these things are resolved.

Fieldworkers visit members in their homes, or a public place such as a café or a library if they prefer. It's a chance for one-on-one time with a professional. Jo Smith, Southern Branch, talks about getting the most from a home visit.

Home visits

Our role is invaluable in that we come into a member's home and see them in their own environment for an hour – you don't get that from many other health professionals.

We're not doctors but we are trained professionals in a

"We're not doctors but we are trained professionals in a health related field, and have a skillset intended to help our members live life to its fullest."

health related field, and have a skillset intended to help our members live life to its fullest.

People welcome us into their own home so it's very important that we establish a rapport with them and meet at a time that suits them. It's their time, they're in charge and we follow their lead.

To begin with, we may ask lots of questions to get an idea of what is important to people, the kinds of challenges they may face and how we might be able to assist. I'm there primarily to listen to what people have to say. Every individual has a story to tell, and by listening to that story we can often work out where we can help most. Then it's a case of prioritising what's most important to them at that moment. It may be issues with pain, or fatigue, or social isolation. Because we work with progressive conditions, it's also important to talk about what may happen next. If you have a plan, often the future is not so daunting.

We are solution focussed and work closely with the member to make sure they are happy with any further steps. Personally, I love being able to leave a member's home knowing there is something we can do that will make their life better. Often it's about exploring options that people may not know exist and facilitating solutions that work for them and their lives. I am constantly inspired by the people I visit.

About half of a fieldworker's time is spent making referrals to other agencies on behalf of a member and supporting them through the process from referral to outcome. Fieldworkers often refer members and advocate on their behalf with various medical and social agencies such as physiotherapy, occupational therapy, needs assessment & service coordination agencies, genetic health services, Housing New Zealand and Work & Income NZ. This is always done with member's consent. Dympna Mulroy, Wellington Branch, explains more.

Referrals

The health system can be difficult to navigate your way around, especially if you are new to it. Some people aren't aware of all the potential supports and services available and it is important to have someone who can walk alongside them on this journey. Letting people know about the support they are eligible to receive and how to access it, enables them more control over their health, and well-being.

Many agencies don't accept self-referrals and some people don't want to go to their GP for a referral given the associated cost, so as trained healthcare professionals we are often able to refer our members to services provided by these agencies. If not, we can advise and help them access it through other means. We are also able to provide accurate and relevant information on a member's rare condition so the agencies are better able to acknowledge and appreciate their needs.

I get lots of positive feedback. I remember helping a member who had been struggling to get in and out of his home because of uneven steps. Previous referrals to occupational therapy had not been successful and he felt the only remaining option was to move. In the meantime he was at risk of falls and injuries. With his consent, I sent a referral to the occupational therapy department requesting they reassess this gentleman's external access and consider a handrail as an alternative means of support. A few months later he rang me to thank me for my support. He explained that the handrail had made a significant difference to his ability to safely access his home.

Unfortunately not all outcomes are positive, especially if someone does not receive the support they hoped for. In these circumstances we support the individual through the process, identify the reason for the outcome and look at what to consider for the future.

A great day at work for me is one when I see someone

achieve a goal. I love seeing someone smile feeling that we have been able to help them have more freedom and choice in their life.

Advocacy

Having someone on your side who can offer support and speak up for you when needed makes all the difference. Advocacy is an important part of the fieldworker's role and Penny Piper from Wellington Branch explains how her job includes everything from going to a WINZ appointment to writing letters to landlords.

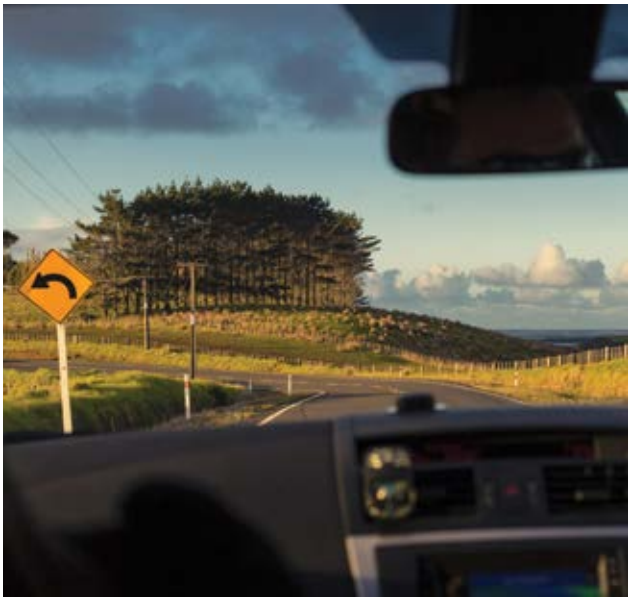
Being an advocate for someone is letting them know that there is someone else walking that road alongside them, who holds their best interests and goals at the forefront.

An advocate can speak up for you if you are feeling a bit overwhelmed and also encourages others to value those with needs different to those of the general population (special housing needs is one example of this). When facing new or challenging situations, having an advocate can also help people deal with feelings of isolation or confusion, supporting them to achieve their desired outcome.

I always talk with our members about what they want and what steps are needed to reach that goal. Then we decide what they would like to do themselves, and what they would like support with.

I may attend WINZ appointments to ensure all entitlements have been discussed, or write letters to landlords, WINZ, and support professionals. Sometimes I help raise issues with carers/agencies, or support conversations with family members that may be difficult. As fieldworkers we support carers and family members who need advocacy too.

Members make comments such as; "I'm so glad to have had help with this", "I'm pleased with the results and next time I would just get on to it sooner", "That's usually been scary for me, but I felt like they heard me when I took a support person", "It was good you reminded me of what I wanted to say".



"When facing new or challenging situations, having an advocate can also help people deal with feelings of isolation or confusion..."

Getting calls or emails that let me know the things we have been working on have been realised is very exciting for me. I get to see firsthand the difference our planning and working together has made.

Regional visits

Our branch offices are based in the main centres but our fieldworkers visit members wherever they live. Much of their time is spent on the road. Marty Price covers Nelson, Golden Bay, Marlborough and the West Coast as far south as Franz Josef and explains what goes into planning a regional visit.

"Some members live in very isolated areas, but one of the great things about our service is that we offer it to everyone, regardless of where they live."

I do a fair bit of planning before heading away, contacting members to organise dates and times and working to see if I can fit all members in on the days I am in the area. I try to see four to five members in a day when away and it can be a juggle at times.

It is important to have time in between visits, for reflecting and preparing, plus having some downtime. Keeping track of documentation and making sure any member information is privately secured whilst at the next visit also takes some managing.

Some members live in very isolated areas, but one of the great things about our service is that we offer it to everyone, regardless of where they live. Some members choose not to have a visit from a fieldworker, which is a personal choice that we respect.

Traveling long distances and in isolated areas means our mobile devices go off the grid and we may drive some distance without seeing other people or towns. Safety and access to professional support is an issue, so keeping in regular contact with our manager or another fieldworker is important.

A lot of members express their appreciation about the service we provide. They appreciate us assisting them with things that they haven't had the resource, support or information to achieve. They also tell me that having someone to speak to who understands their condition is really important and they are glad they can get this from their fieldworker. Members want this level of understanding from their GPs and hospital staff, but don't always get it.

It's a good day when I can walk back to my car still seeing the smile on the face of the member I just visited. During our visits, I find people just release their thoughts and frustrations and sometimes we come up with a plan of what to do next. But even if there's no plan to follow up, it's good to know I was there to listen.

Professional development

While fieldworkers work alone a lot of the time, they are part of a team and check in with each other regularly to maintain best practice and keep up to date with new developments in research and treatment. Paul Graham, Canterbury Branch, explains how.

It is important to check in with other fieldworkers, either on a one-on-one basis, or via our regular teleconferences. I hear what is going on in the other regions, and can run questions and issues past others, who may have ideas on what to be mindful of with a particular condition or how to best approach a particular problem. These catch-ups are rather like working in the same office. It is very helpful to be able to talk to another fieldworker and compare notes. It can be a relief to know that I am not working alone and also good to get acknowledgment for the things that I am doing well. I can also contribute to, and share in other's learning, which is great.

We all attend training sessions at National Office a few times a year and we each maintain a professional membership with an affiliated professional body, depending on our base training and discipline. Our team is proactive about professional development.

The MDANZ Fieldworker service is offered free of charge to MDANZ members and is funded through donations and grants. Contact your local MDANZ Branch to be put in contact with your fieldworker.

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Stronger together

Reaching out and fitting in

Beat stress and find a sense of belonging with a strong social support network

Talitha's story

For 17 long years following her diagnosis, Talitha Vandenberg felt very much alone. Her doctors answered as many of her questions about myasthenia gravis (MG) as they could, but often their answers felt like they came from a text book, rather than real life.

"I barely met anyone else with MG and spent all my time telling people all about it, so at times felt more like an educator than someone with the condition," she says. "I felt very alone and not understood. No one had heard of it, and although people were nice and tried to understand, sometimes I had to give up because they couldn't understand the gravity of MG and the effect it has. So, over time I internalised my loneliness."

But several years ago, Talitha decided to reach out and find others with the same condition, hoping to put an end to both her loneliness and theirs. Although she had a close network of family and friends around her, she wanted to get in touch with people who shared her condition. She created a website, told her story to the local papers and asked people to make contact. Talitha found that people were travelling significant distances to meet her and others with MG in Pahiatua.

"I have noticed that given half a chance, people will take great steps to finally have a moment of inclusion." Talitha applied to MDANZ's Discretionary Fund and was pleased when instead of simply granting her funds to meet with others in Christchurch, they committed to working together to facilitate these groups.

Some travel from Auckland, and many drive at least two or three hours for the chance to share information, strategies to make life easier, and – just as importantly – to have a "good old-fashioned chat" over hot drinks and snacks.

"I have been told that attending the group has been life-changing," says Talitha. "All the feedback I receive has been resoundingly positive. Most people say they can't wait for the next one."

It's also meant a lot for Talitha personally.

"It has given me a sense of purpose and inclusion. I'm not living in the shadow of MG now, MG lives in the shadow of me. It has given me self-confidence and improved my self-esteem. I have made some wonderful friends.'

In between meetings, the group keeps in touch via Facebook. Following on from a visit by Talitha, a group now gets together in Christchurch, and there are many plans for further meetings. If you have MG and would like to connect with others visit Myasthenia Support Group, New Zealand on Facebook.

Why get together?

Many studies show that strong social support systems are one of the major factors in psychological well-being and physical good health.

Support groups can be formal or informal, and vary in size and structure. In general, a support group is a place where people are able to talk with others who are going through similar things and so truly understand the pressures they are facing and can share practical insights that can only come from firsthand experience.

Talking to others and / or doing things together in support groups reduces anxiety, improves self-esteem and helps with overall sense of well-being. People living with a neuromuscular condition often experience high stress levels, as well as isolation. Building and maintaining a strong social network is a proven way to becoming more resilient and offers some comfort in knowing you have access to friendly support, information and advice when times get tough.

And with mobile technology becoming more popular and accessible, support doesn't have to mean face-to-face contact either. Making friends online and joining internet-based support groups can be just as effective, especially if there is nothing on offer where you live.

Online support

When Alex Heke's son Tana was diagnosed with Duchenne muscular dystrophy (DMD), Alex and her family hadn't even heard of the condition. She was very keen to get in contact with other families but couldn't find anything local on facebook so she started the Kiwi Kids with DMD page on Facebook.

"When our son was diagnosed I felt very alone and scared," says Alex. "I felt as though we were the only family going through such a terrible time but since starting the page we have formed a great little group. Some of our members joined when they had a new diagnosis. It was great they were able to have the support of other newly diagnosed families, and those of us who had already been through it all. It is something I wish we had when Tana was first diagnosed."

Members of the page are a supportive and friendly group of parents, grandparents, and friends, along with boys and men with DMD. It's a closed group so comments and discussions can't be seen by anyone who doesn't belong to the group.

"The page gives ourselves and other parents a chance to be able to chat and vent to each other about everyday things that we deal with when it comes to DMD," says Alex. "I find it a lot easier to talk to a parent that knows what we are going through. It is great to be able to exchange ideas and opinions amongst each other and even better being able to meet other families with the same condition as our son. It makes dealing with DMD that little bit easier."

I felt as though we were the only family going through such a terrible time but since starting the page we have formed a great little group.

There are a number of international groups for families living with DMD, but treatments and medications differ from country to country, and a local group has the added potential of being able to meet other families in person.

"I am happy to say I have made really good friends through the page,' says Alex.

We can help

One of MDANZ's new organisational values is Connected (*Tūhonotanga*). You'll find a list of support groups on our website www.mda.org.nz/information/support-groups/ and if you want to set up a new group, we can help out with suggestions and guidelines. If you are interested in getting more connected, the best person to talk to is your fieldworker.

Choose your own direction

Challenge yourself and make new friends

The world's leading youth achievement award
is now available through MDANZ.

For the last 60 years, millions of young people all over the world have had the chance to experience challenge and adventure, acquire new skills and confidence, and make new friends – all thanks to the Duke of Edinburgh Award, known locally as the Duke of Edinburgh Hillary Award.

We're delighted our young members will be able to experience the same things with MDANZ becoming a licensed Award Unit.

Chief Executive Ronelle Baker says; "It aligns itself strongly with the values of our organisation of empowerment, being connected, proactive and sustainable – creating opportunities for our young people living with neuromuscular conditions to live a life of freedom beyond limits, to build leadership capabilities and relationships within their communities and beyond, assisting us to influence social change and limiting beliefs placed on people with impairments in our society."

The award, with its instantly recognisable levels of Bronze, Silver and Gold, is voluntary, non-competitive and available to anyone aged 14-24. It is all about individual challenge. Whether you are wanting to gain new life skills and confidence, or adding to your CV and employment chances, this is a great programme to be a part of.

Fieldworker Marty Price is our award leader. Marty is based in Nelson and has worked in the disability sector for 27 years. He is passionate for people about equal rights and community inclusion. As a coach and player of wheelchair basketball, Marty works with a number of young people and is looking forward to being a mentor and coach to our "Dukies" – Duke of Edinburgh's Hillary Award participants.

His goal is to support every "Dukie" to tailor the programme to their individual needs and achieve their goals.

"The only person they compete against is themselves, by challenging their own beliefs about what they can achieve," says Marty.

What are the benefits?

- The Award could play a vital role in providing opportunities for young people to develop essential life skills, increase their employability and foster their creativity, and innovation.
- It can help young people become more socially confident, helping them to believe in themselves and their abilities, rather than focusing on their disability.
- The Duke of Edinburgh's Hillary Award is a prestigious and well recognised award, highly regarded by employers as it shows determination, commitment, leadership and perseverance.

What is involved?

Young people design their own award programme, set their own goals and record their own progress.

They choose a:

- Service: For young people who often have to rely on the assistance of others, the ability to contribute and give service is highly empowering and great for sense of self-worth
- Physical Recreation: This can be as much or as little as an individual is capable of stretching themselves to achieve
- Skills activity: This could be an existing skill or something new such as gardening, fishing, baking etc
- Adventurous Journey: This involves planning and complete a trip away from home
- Residential Project: This is an additional live-in programme completed at Gold level

A small budget will be allocated at each level of Bronze, Silver and Gold, to help each participant to achieve their goals.

To complete the Bronze Award, a commitment to the skills, service and physical recreation sections requires three months for two sections and six months for one section. For example, you may decide to volunteer (service) for six months, learn umpiring (skills) for three months, and play soccer, wheelchair or otherwise (physical recreation) for three months.

The time commitment for each section is to perform the task for a minimum of one hour a week. The Adventurous Journey for a bronze award, is a two day/one night trip. You can take as long as you like to complete each level. You may decide to complete only one level or all three levels up to Gold.

How do we know this will succeed?

Muscular Dystrophy NSW has been running this programme for their young people for five years.

They love that The Duke of Edinburgh's International Award is not a special programme or modified for people with a disability. Instead, it provides an opportunity that is the same for all young people.

National Office hosted "Dukie", Scott Green, and MD NSW Duke of Edinburgh Coordinator Melissa Wentworth-Perry last year. Scott was very young when he was diagnosed with Duchenne Muscular Dystrophy (DMD), but has not let it slow him down. He became one of the 'faces' of MDNSW in order to fulfill his aim of raising awareness and funds.

He began the Duke of Edinburgh's Award, in early 2014 and is still working on it. Scott has learnt new skills, challenged himself and exceeded the expectations of those around him. Scott is now an ambassador for Muscular Dystrophy NSW and NSW Kids in Need.

"It has changed my life as it has given me something other than myself to think about and made me contemplate future possibilities in terms of work and hobbies," he says.

To see more about the Muscular Dystrophy NSW experience go to: mdnsw.org.au/the-duke-of-ed-overview/

Whether you are looking for friendship, new skills, improved abilities, excitement, wider horizons, confidence or developing connections locally, nationally and internationally, it's all available in the Duke of Edinburgh's Hillary Award. Award Leader Marty would love to talk to our young members or their parents about starting the journey. Please call him on 027 4446681 or email Marty@mda.org.nz

First approved treatment for spinal muscular atrophy!

Nusinursen approved by FDA in the US.

Just before Christmas, the United States Food and Drug Administration (FDA) announced that it has approved Biogen's Spinraza (known as nusinursen in other parts of the world) as a treatment for SMA. This approval is for a broad licence across all types of SMA, which means that nusinursen can be marketed as Spinraza in the US for children and adults with all types of SMA.

In ENDEAR, a pivotal controlled clinical study, infantile-onset SMA patients treated with nusinursen achieved and sustained clinically meaningful improvement in motor function compared to untreated study participants. In addition, a greater percentage of patients on nusinursen survived compared to untreated patients. In open-label studies, some patients achieved milestones such as ability to sit unassisted, stand or walk when they would otherwise be unexpected to do so and maintained milestones at ages when they would be expected to be lost. The overall findings of these studies support the effectiveness of nusinursen across the range of SMA patients, and appear to support the early initiation of treatment.

Biogen is committed to working with New Zealand's government agencies to bring nusinursen to Kiwis with SMA. MDANZ is supporting Biogen in its efforts as required so its members with SMA can benefit from this treatment as soon as possible.

First reported by Bloomberg News, Spinraza will be one of the costliest medicines in the world at US $125 000 per dose. During its first year of use six doses are required with three doses per year after that. The drug's high price is consistent with other treatments for rare diseases for which there are no other options and it is likely that if there were competing therapies there would be pressure to decrease the price. The drug's delivery method, intrathecal (spinal) injection is also costly adding to the overall expense of the treatment. R

DMD treatment to continue

The treatment of Duchenne muscular dystrophy caused by a nonsense mutation (nmDMD) with Translarna will continue in Europe. The European Commission authorised the renewal based on a continued positive benefit and risk assessment. The renewal allows PTC Therapeutics to continue to market Translarna for the treatment of nonsense mutation Duchenne muscular dystrophy (nmDMD) in ambulatory patients aged five years and older in the 28 countries that are Member States of the European Union, as well as European Economic Area members Iceland, Liechtenstein and Norway. As a specific obligation of the renewal, PTC will conduct an additional trial of Translarna which may involve Australian and New Zealand boys whose DMD is caused by a nonsense mutation. R

New clinical trial

A clinical trial for people with myotonic dystrophy type 1 (DM1) is taking place at the John Walton Research Centre in Newcastle, UK, sponsored by AMO Pharma. The study will investigate the safety of tideglusib (AMO-02), a drug that inhibits an enzyme called glycogen synthase kinase 3ß (GSK3ß), over-active in people with DM1. Preclinical research has shown inhibiting GSK3ß can increase muscle strength and decrease myotonia in mouse models. The study will also investigate the efficacy and the pharmacokinetics of tideglusib. R

Orphan Drug designations for SMA and Oculopharyngeal muscular dystrophy

Understanding the process in Europe and the USA.

In January this year the U.S. Food and Drug Administration (FDA) granted orphan drug designation to RG7916 for the treatment of patients with spinal muscular atrophy (SMA).

RG7916 is part of PTC Therapeutics' joint development programme with Roche and the US-based SMA Foundation.

There are two clinical studies: SUNFISH, a trial in childhood onset (Type 2/3) SMA patients, and FIREFISH, a trial in infant onset (Type 1) patients.

Spinal muscular atrophy (SMA) is a rare genetic disorder that results in disability beginning in infancy and is the leading inherited cause of mortality in infants and young children.

In January this year another biopharma company Benitec Biopharma announced that the European Commission (EC), based on a favourable recommendation from the European Medicines Agency (EMA) Committee for Orphan Medicinal Products, has granted Orphan Drug Designation to BB-301 for the treatment of patients with oculopharyngeal muscular dystrophy (OPMD).

OPMD is a rare inherited myopathy characterised by dysphagia (difficulty in swallowing), the loss of muscle strength, and weakness in multiple parts of the body. OPMD is not typically diagnosed until the

individuals reach their 50's or 60's. As the dysphagia becomes more severe, patients become malnourished, lose significant weight, become dehydrated and suffer from repeated incidents of aspiration pneumonia. The latter two ailments can result in death. Currently, therapeutic strategies employ repetitive surgical interventions that have limited efficacy.

BB-301 is a genetic therapeutic for the treatment of OPMD comprised of a single expression construct for the 'knockdown and replace strategy' of mutant PABPN1, the main cellular component that leads to OPMD in humans. BB-301 is currently in preclinical development and Benitec plans to initiate further studies later this year. Entry into the clinic with a Phase I/II study in OPMD patients is anticipated in 2018, subject to toxicity results and future regulatory review.

In the U.S., orphan drug designation is granted by the FDA's Office of Orphan Products Development to promote the development of products that may offer therapeutic benefits for rare diseases. Orphan drug designation provides opportunities for grant funding towards clinical trial costs, tax advantages, FDA user-fee benefits, and seven years of market exclusivity in the United States, if granted FDA approval.

Similarly, orphan drug designation by the EC provides regulatory and financial incentives for companies to develop and market therapies that treat a life-threatening or chronically debilitating condition affecting no more than five in 10,000 persons in the EU, and where no satisfactory treatment is available. In addition to a 10-year period of marketing exclusivity in the EU after product approval, orphan drug designation provides incentives for companies seeking protocol assistance from the EMA during the product development phase, and direct access to the centralised authorisation procedure.

Orphan drug designation in one jurisdiction does not mean that orphan drug designation will automatically follow in another jurisdiction.

Rare disorders take centre stage

February 28th is global Rare Disease Day – this year's theme is Research.

Research is key. It brings hope to the millions of people living with a rare disorder across the world and their families, including families living with rare neuromuscular conditions in New Zealand.

Although people with lived experience of rare disorders face many challenges, enormous progress is being made every day.

The ongoing implementation of a better comprehensive approach to rare conditions has led to the development of appropriate public health policies in some countries. Important gains continue to be made with the increase of international cooperation in the field of clinical and scientific research, such as that implemented by TREAT NMD, as well as the sharing of scientific knowledge about all rare conditions. These advances have led to the development of new diagnostic and therapeutic procedures. See Michelle's story on page 26 to read a first-hand account of how research has positively changed the life of someone living with a rare condition. ®

www.rarediseaseday.org

Deflazacort approved in the USA

In February 2017 the US Food and Drug Administration (FDA) approved deflazacort for the treatment of Duchenne in children aged five years and older. Marathon Pharmaceuticals conducted numerous new preclinical and clinical studies to support the New Drug Application to the FDA. This investment in research and development has led to new understanding of the dosing, drug interaction and safe use of deflazacort. Additional research will continue to advance the science of the drug and care of patients. One planned study will examine dosage regimens in younger patients to determine if earlier intervention is safe and effective and ultimately impacts the course of the disease. A second study will examine dosage regimens in non-ambulatory patients and characterize the pulmonary and cardiac effects. ®

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Congenital Myasthenic Syndrome (CMS)

What is Congenital Myasthenic Syndrome?

Congenital Myasthenic Syndrome (CMS) is a family of inherited neuromuscular conditions characterised by skeletal muscle weakness that worsens with physical exertion. Cardiac and smooth muscle are usually not involved. Coordination, sensation, and tendon reflexes are normal as are cognitive skills. Myasthenia (muscle weakness) is due to problems in the neuromuscular junction, which is the area between the ends of nerve cells and muscle cells where signals are relayed to trigger muscle movement. Symptoms of muscle weakness typically begin in early childhood, however they can also begin in adolescence and adulthood. The severity of the myasthenia varies greatly, with some people experiencing minor weakness and others having such severe weakness that they are unable to walk. Prevalence of CMS is unknown.

What are the different types of Congenital Myasthenic Syndrome?

The types of CMS are grouped into three main categories depending on the part of the neuromuscular junction affected. These include presynaptic (the nerve cell), postsynaptic (the muscle cell) or synaptic (the space in between the nerve and the muscle cell).

Presynaptic CMS is characterised by insufficient release of acetylcholine, a neurotransmitter that controls muscle contractions. This affects 7-8% of individuals with CMS.

The features of CMS are often present at birth; however sometimes symptoms are not noticeable until adolescence.

Postsynaptic CMS have two forms, and affect a total of approximately 75-80% of individuals with the condition. One is characterised by missing acetylcholine receptors or receptors that don't stay open long enough called "fast-channel CMS" and the second is characterised by acetylcholine receptors that are open for too long, called "slow-channel CMS".

Synaptic CMS is characterised by a deficiency of acetylcholinesterase, an enzyme which breaks down acetylcholine. This affects 14-15% of individuals with the condition. Identification of the specific subtype is important in patient care for determining the most effective treatment.

What are the features of CMS?

The features of CMS are often present at birth; however sometimes symptoms are not noticeable until adolescence. Infants with CMS may be slow to meet their crawling or walking milestones and children with CMS have visible facial weakness. Affected infants can also have periods of shallow breathing often at times of infection, fever or excitement which can cause cyanosis (blue skin or lips). An abnormal, high-pitched, musical breathing sound caused by a blockage in the throat or voice box (larynx) can also be heard in CMS infants when taking in a breath. Other symptoms of CMS are difficulties with chewing, swallowing or feeding and choking spells due to muscle weakness of the mouth and throat. The eye muscles are also commonly affected causing droopy eyelids (ptosis). Curvature of the spine (scoliosis) and in some cases joint contractures can be present.

What causes CMS?

CMS is an inherited neuromuscular condition. It is caused by alterations in genes necessary for making the acetylcholine receptor or other components or proteins of the neuromuscular junction. Mutations in the *CHRNE* gene are responsible for more than half of all cases. A large

number of cases are also caused by mutations in the *RAPSN*, *CHAT*, *COLQ*, and *DOK7* genes. Except for slow-channel CMS, the inheritance pattern for the different types of CMS is autosomal recessive. This means that it takes two copies of the defective gene, one from each parent, for the disease to be present. Slow-channel CMS is autosomal dominant; therefore, it only takes one copy of the gene from one parent to cause the disease which means that there is a 50% chance of an affected parent to pass on the disease to their child.

Diagnosis of Congenital Myasthenic Syndrome

A full comprehensive family history and physical examination is part of the diagnostic process. The physician, usually a neurologist, will be looking specifically for weakness and fatigue, particularly in response to physical exertion. The strength of eyelids and skeletal muscles may be assessed by asking the patient to look towards the ceiling without blinking and holding their arms out for as long as possible.

If physical tests are consistent with myasthenia, blood tests will be ordered to detect antibodies to the acetylcholine receptor. A negative test will rule out myasthenia gravis (MG), an autoimmune disease and may indicate possible CMS. However, it does not rule out seronegative types of MG. Electrodiagnostic tests, where an electrode is placed on the surface of a major muscle and a small shock delivered to the nerve, record the responses in the muscle to contraction.

An intravenous injection of Tensilon, a fast-acting acetylcholinesterase inhibitor may be administered as part of the diagnostic process and a temporary increase in strength after the injection would indicate CMS.

Certain drugs should be avoided by people with CMS as they are known to affect neuromuscular transmission and exacerbate symptoms of the condition ...

Muscle biopsy and the absence of major pathological findings as well as genetic testing can further conclude the type of CMS and a family history of myasthenic syndrome would support this diagnosis.

Management of CMS

Non-invasive ventilation at night to help with breathing difficulties. Apnea monitors are recommended for young children.

Treatment with medications is available for many types of CMS;

- **Pyridostigmine**, a cholinesterase inhibitor, enables messages to travel from the nerve to the muscle. This is used for presynaptic CMS and postsynaptic fast-channel CMS.
- **3,4-diamino-pyridine' DAP**, increases acetylcholine release which causes electrical messages to last longer. This is used in postsynaptic fast-channel CMS.
- **Ephedrine and/or albuterol (salbutamol)** can improve muscle strength.
- **Quinidine or fluoxetine** to help faulty acetylcholine receptors to close for post synaptic slow-channel CMS.

There are no medications currently available to treat synaptic CMS.

Certain drugs should be avoided by people with CMS as they are known to affect neuromuscular transmission and exacerbate symptoms of the condition and include ciprofloxacin, chloroquine, procaine, lithium, phenytoin, beta-blockers, procainamide, quinidine.

Research into Congenital Myasthenic Syndrome

Current research studies are focusing on the development of a new mouse model of a CMS; genetic analysis of a worm model of a slow-channel myasthenic syndrome; studies of how the nerve-muscle function forms and also methods to improve diagnosis, treatment and prevention of congenital myasthenic syndrome.

The power of perseverance

Michelle Crane's journey to diagnosis was long, but life-changing

"I was born, the youngest of four, to very loving caring parents. I suffered from breathing difficulties and was transferred to National Women's Hospital and then to the Critical Care Unit at Auckland Hospital where I remained for eight months.

I required repeated intubation, naso-gastric tube feeding and a gastrostomy was performed at one month of age and continued for seven months. At age 9 months I weighed 6lb 3oz – only one ounce more than when I was born. At 18 months of age I was finally discharged from Auckland Hospital and allowed home for the first time.

Between the ages of two years and four years I reached normal milestones but my parents noticed I was slow in getting around and playing. By the age five I had a really noticeable sway back. My mum took me to the GP, who told her "not to worry, she has a sway back, she'll outgrow it". From the age of 6 I found school very difficult as I could not walk far, would tire easily and at one point was found crawling to the bus as I was so weak.

Michelle with Jacob when he was three-months old.

I was taken to chiropractors for treatment, which I found painful. I had many exercises to do at home including one where I would have a brick tied to my foot and walk with it in the hope that this would improve strength in my legs. As time went on and my parents could see no improvement I was taken back to the GP who referred me to specialists at Auckland Hospital. After a range of tests, including a muscle biopsy it was concluded I most likely had limb girdle muscular dystrophy, although they could not be certain. I was about 11 years old and I remember we were told there was nothing that could be done, I would most likely be in a wheelchair by the time I was in my early teens and my life expectancy was unsure. I remember feeling very alone, like I was the only person in the world with my condition and somehow I just had to deal with it.

After my diagnosis there was never any follow up appointments or support for me or my parents. Life went back to normal. Mum and Dad didn't treat me any different from my two older brothers and older sister and always encouraged me to be as independent as possible. As there was such a huge gap between myself and my siblings it wasn't long before I was the only one left at home. As I didn't really have any friends at school because of my disability, the kids just didn't understand, I grew up very lonely and, at times, very depressed. But I was determined to make the best of my situation. I went to high school but by the time I was 16 I couldn't cope with getting around and as I didn't have a wheelchair I left school and did 6th form by correspondence.

I felt I needed to understand more, I started questioning my life expectancy, my future and whether I was going to be there for my son.

My parents continued to be a huge support for me growing up. At age 18 I left home and went flatting by myself. I had an office job and a car and was as independent as I could be. When I was 26 years old I fell pregnant with my son Jacob. I found the pregnancy very tough and had to leave my job as I was just too exhausted physically to do much.

I started wondering again about my condition, and when my son was born the realisation that I couldn't even lift him, made me want to seek specialist help again. I felt I needed to understand more, I started questioning my life expectancy, my

future and whether I was going to be there for my son. I also wondered if there was a chance he would have the same condition or would he pass it on. He was the real driving force behind me wanting to seek help and understanding and i wanted to get better and stronger for him.

Through my GP I got an appointment with Neurology at Auckland Hospital and so the process of determining exactly what I had bagan again. Once again I had extensive testing, including another muscle biopsy, but alas to no avail. I just didn't really fit into any category. By this time I had become extremely weak and when my son was about 6 or 7 I had to start using a C-Pap machine at night for breathing as it was too difficult to breathe lying down. Every year I went back to see my neurologist, Dr Richard Roxburgh, for a check-up and testing for different types of muscular dystrophy. At one appointment he discussed gene sequencing research and asked me if I would like to participate. I agreed wholeheartedly as I had nothing to lose and so a sample of my DNA was sent overseas.

After 18 months of waiting I received a call to come in and see Dr Roxburgh at the hospital. It was the day before Christmas. The result had come in and showed that I had Congenital Myasthenic Syndrome. I could take a drug, Salbutamol which could significantly help me with my symptoms. It had been over 30 years after I first presented with symptoms and sought diagnosis. This was the best Christmas present ever. I cried all the way home. Finally I had an answer. It felt like a huge relief, not just to me but for my parents and my son.

This was the best Christmas present ever. I cried all the way home.

Since then life has been even more wonderful than I could imagine. I have been able to do so much more physically than I have ever been able to do. Yes, I still need to use my wheelchair for any long distance walking but the small everyday things like getting dressed, making a coffee and showering have become so much easier. I feel for the first time in my life "normal".

I was given a lot of information and support this time, I felt I could finally understand what's going on, and that has given me a lot of hope for the future, knowing finally that I was going to be around for my son, and that I was going to be able to do things like walk again. I was also reassured that it was highly unlikely to be passed on, I was the only family member to have it.

I currently feel a lot stronger, I was referred to physio to improve my strength and I am on only one medication, Salbutamol, which has made a huge difference to my strength and my breathing.

My son Jacob, now 16, is a huge support for me. I am very proud of him, growing up with a mum that wasn't able to do much has taught him to be empathetic towards other people. He is my biggest and proudest achievement in life and I have high hopes for him.

Top: A nurse holds a 10-month-old Michelle
Above: Michelle aged 7 with her pet lamb

If it wasn't for my parents making me such an independent and stubborn person I don't think I would be where I am today. I always knew in my heart that I would get answers and with lots of persistence and perseverance it paid off. My parents instilled in me a desire to not ever give up and that's my advice for anyone else; keep thinking positive, stay strong and believe."

To read more about Michelle Crane's diagnosis, read Richard Roxburgh's column on page 30.

GUEST speakers

The power of research

DR. RICHARD ROXBURGH

Lives are changes when an international community works together.

I hope you've enjoyed reading Michelle's story of what it's like to live with congenital myasthenic syndrome on page 26. It is likely that Michelle's life would have followed a different path if she had been accurately diagnosed as an infant. Her condition is rare and even now it would be hard to make a diagnosis based just on the clinical features but nowadays patients like this who are baffling, can have a panel of gene tests and sometimes as in her case, once the genetic result comes in the whole clinical picture falls into place. Use of these genetic panels is becoming much more widespread and where used well will definitely shorten the time to diagnosis (and treatment where it's available) to months – rather than more than 30 years!

The advances in knowledge about genetic diagnosis and management of neuromuscular conditions don't just happen by themselves but occur due to the efforts of many people involved in research. Michelle received her long-awaited diagnosis of congenital myasthenic syndrome after enrolling in one of our research studies which was part-funded by the MDANZ's research trust, Neuromuscular Research New Zealand.

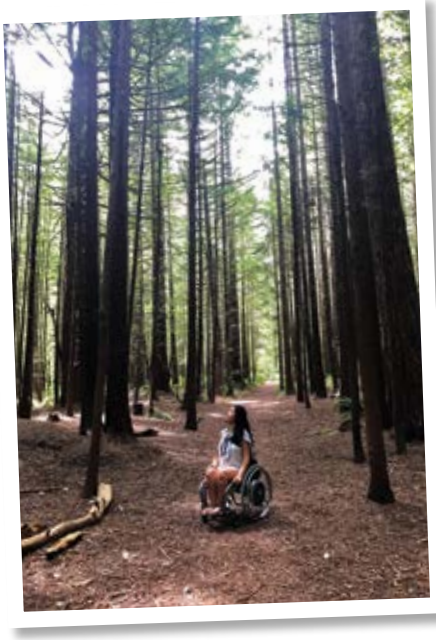
In May 2014, when I was attending the annual Australasian Neuromuscular Network meeting in Melbourne, Nigel Clarke, a fellow New Zealander working in Sydney at Westmead Children's Hospital's Institute for Neuromuscular Research (INMR), generously offered a number of places for New Zealand patients on a study that one of his PhD students, Dr Roula Ghaoui, was conducting. Roula's research involved using Next Generation Sequencing to find the genetic cause of limb-girdle muscular dystrophy in previously undiagnosed patients.

Miriam Rodrigues and I contacted more than 30 patients, explaining to them the nature of the study; the possible benefits namely finding the genetic cause of their condition and the possible risks around finding genetic mutations causing something other than muscle weakness and finding genetic variations of unknown significance. Over the following months Michelle, along with 27 other patients from New Zealand agreed to take part in the study.

DNA samples were sent to Sydney and then on to Prof Daniel MacArthur's lab at The Broad Institute at Harvard in the USA, where a technique called Whole Exome Sequencing was performed which basically looks at all the genes at once. Subsequent analysis by Roula Ghaoui and Sandra Cooper at the INMR found that Michelle had an autosomal recessive condition caused by mutations in the DOK-7 gene. Don Love's clinical lab in Auckland confirmed the research result and issued their report back to me on 21st December 2015. Two days later I was able to meet with Michelle to discuss with her this long awaited diagnosis. It was exciting to read that the type of congenital myasthenic syndrome (which was first discovered by American researchers in 2006) had been shown in another research study by Brazilian and British researchers, in 2013, to be treatable with a readily available asthma medication – and one of the highlights of my career to date - when she came back to clinic without her wheelchair! Furthermore, this result has also relieved Michelle and her family members of their anxieties over the possibility of her passing the condition on to her son.

I am frequently asked what the point of genetic research is – patients get a diagnosis – so what? Well, if any of the researchers either here in New Zealand, or in Australia, USA, Brazil or UK hadn't played their part then Michelle would still be in a wheelchair.

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



A change of scenery makes all the difference.

Enjoying the moment

OLIVIA SHIVAS

Happy 2017 to all – it's crazy to think another year has gone by (and that we are already a few months into another one!)

Over the summer I spent a lot of time catching up with friends and family, trying out new cafes and going on bush walks. Holidays are also a really nice time to pause and relax; there's no need to stress about work and other commitments.

One of the highlights from my holiday was visiting family in Tauranga. We walked around the base of Mt Maunganui, which was a little steep in some places but definitely suitable for wheelchair-users. The views from the Mount are so beautiful and it made me really appreciate living in New Zealand.

Starting a new year is an interesting time – you reflect on the past and year that's been, but you also think about the future and having a fresh start.

Starting back at work was a little bit hard after all the sleep ins and time in the sun, but as a friend reminded me – I should be thankful I have a job I enjoy to go to every day!

Starting a new year is an interesting time – you reflect on the past and year that's been, but you also think about the future and having a fresh start.

I'm not really one to write down a list of specific resolutions, but each year I do like to find new ways to make life more fulfilling and interesting. This year I want to focus on being present and enjoy the moment I'm in.

I want to go on more adventures with friends and stop making excuses for not doing things. I've already booked some holiday homes for weekends away throughout the year with family and planned some trips with friends. My weekly schedule is pretty busy with work, but I think it's important to allocate time to take a break and refresh – even if I have to plan it months ahead!

I've also made a pact with some friends that we will go on a walk (well, I'll roll!) once a week. Not only does it mean I can regularly catch up with them and work in some exercise, but getting some fresh air is really good for your mental health. I'm also excited to discover new places to explore in Auckland and have a change in scenery.

Taking time to reflect.

I'd love to hear from other young MDANZ members and know what you got up to over the holidays. You can email me at olivia.shivas@gmail.com to get in touch.

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.

Ask the **PANEL**

Seeking change and enjoying life

DR HUHANA HICKEY

Q: I recently booked tickets online to take my daughter to the movies. When we got to the theatre, the lift was out of order and there was no way I could get into the theatre. While they were happy to refund my ticket, our evening out was pretty much spoilt. What responsibilities do commercial premises have not just to make sure their premises are accessible, but that they are kept in working order. Should their website have included information or let me know the lift wasn't working?

When those of us with disabilities head out into town – or anywhere at all – we can't be spontaneous. We need to know what the access will be like at the places we visit.

When I went to Waitangi over Christmas I refused to pay the requested price as there was no guarantee I could actually access the grounds in a wheelchair because of uneven footpaths and steep gradients. I challenged it, and got in for free so I could check it out. My concerns were confirmed and I gave the manager some advice about improving access so they could justify their entrance charges.

This is a common experience for many of us. As far as this question is concerned, why did the website not forewarn people with disabilities that the lift was out of order?

There is no rule that says they must notify patrons beforehand and that's the problem. Disability access is such a low priority for many retailers and businesses that we are often just not considered. However, I suggest you make your displeasure known at the time, and even contact Consumer Affairs. While they may not be able to do much, it can highlight an issue they may wish to take up.

They can be reached at: https://www.consumerprotection.govt.nz/report-or-resolve-a-problem/general-problems/

The other thing that can be done is to arrange a meeting with the manager. It might be a good idea for a group of people representing several disability organisations to band together and ask the manager to change the way the venue communicates any barriers a customer with disabilities may experience. It is an opportunity to highlight the need to put this info on the website and at the front of their building with a phone number or email for people to contact.

If this is a recurring issue, then it might need to go further and there are a range of options for that such as the Human Rights Commission, although it will be seen as a goods and service issue. It can also be useful to go to the media, as sometimes change only occurs when the issue is stirred up. The bottom line is you do not have to accept the situation and therefore you have a right to demand service. If they refuse to make it accessible and create discriminatory barriers, then it becomes a human rights issue. Unfortunately we don't have great building codes around access. The commercial sector only does the bare minimum and so we have to keep plugging away until they can get it right and realise we are citizens with money to spend to enjoy ourselves. So demand access, seek change and go out and enjoy life. Do not let these little issues hold you back.

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.

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Mark "Wheelchair Junkie" Smith

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T. Gross I love mine as well. I've been truly blessed with this wheelchair!

M. Henson I'm just weeks away from getting this chair and I can't wait. I've had an opportunity to use a demo for a week and this chair is a game changer.

D. Heilman Flipping fantastic! Everyone deserves to look at another eye-to-eye... not up or down.

L. Boccuti This is so great. My husband felt claustrophobic in his chair in a crowd because he was so far down. This would have helped that so much.

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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 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

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Wellington Branch

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Canterbury Branch

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Southern Branch

Fieldworker: *Jo Smith*
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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

Peter Tegg. Ph: 0272462145
Email: Peter.Tegg@wcc.govt.nz

Southern Branch

Raewyn Hodgson. Ph: 03 486 2066
Email: raewyn.hodgson@xtra.co.nz

Canterbury Branch

Warren Hall. Ph: 03 329 4390
Email: warrnjh@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

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