InTouch

KIA NOHO TATA Inclusive | Inspiring | Informative Spring 21 Issue 111

Ready, set, roll

Neil is ready to roll for our annual appeal

Yes, you can…

Taking inclusive travel to the next level

Young, strong and independent

Brittney is giving back to the community

Muscular Dystrophy New Zealand

InTouch

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Kōrero with Trevor

Working through the challenges.

The past few months have seen New Zealander's resilience challenged; with wild weather hitting our Southern Region, the trans-Tasman bubble affected by a Covid-19 outbreak and the usual ills and chills winter brings.

However, Kiwis are known for their strength and determination to get through challenging times and none more so than Jezza Williams. His story is inspirational, and I hope you find it as motivating as I did. Jezza set himself a big challenge – making the adventure tourism industry accessible – and won (page 12). With only a few months left in the year, I encourage you to set yourself a challenge too!

Our annual appeal is coming up this month and it's an incredibly exciting new fundraising campaign called Roll 1k for MDA.

The challenge is to complete 1km in distance using any kind of wheels, raising awareness and vital funds for MDANZ along the way.

This is a fun and inclusive campaign that all your family, friends and colleagues can get behind. I encourage you to sign up, spread the word and get creative with the wheels you use. This is the perfect opportunity to challenge YOU.

Challenges in life can be difficult to overcome, whether physically or mentally, but can be done with willpower, determination, and support.

Support is one area where MDANZ can assist you and your whanau through our Fieldwork service.

Our team of Fieldworkers provide personalised support and education right from diagnosis. Even if you have been a member for many years, please do not hesitate to reach out to your Fieldworker for assistance.

I am excited to announce a new Fieldworker has started in the Northern Region, Michael Schneider. You can read more about him on page 3.

MDANZ can also support you via the Bradley Jenkin Memorial Fund which provides discretionary grants for members to access opportunities and specialised resources that enable them to achieve or overcome their challenges. Daniel Lyall, a member from Christchurch, shares his story on page 18 of how the grant has helped him design and produce products using a 3D printer to help himself and other members.

I hope you enjoy the warmer days spring brings and I look forward to hearing all about your challenges for the Roll 1k for MDA fundraiser.

Mauria te pono – believe in yourself.

Trevor Jenkin
National Executive Chairperson

MDANZ staff on the move

This edition we welcome a new staff member and a returning team member to the MDANZ family while we farewell another.

Mike Schneider has joined the team at the Northern branch as a Fieldworker. Mike is no stranger to MDANZ – he and his family have been members of the organisation since their son was diagnosed with Duchenne muscular dystrophy in 2011.

Mike joined the Northern branch committee a few years later where he has held the roles of committee member, branch representative on the National Council and branch Vice-Chair.

Prior to joining MDANZ as a staff member in July, Mike was working as a Community Support Coordinator for CCS Disability Action.

He is looking forward to meeting members and being available to assist them however he can. We're delighted to have Mike join our team.

MDANZ is pleased to welcome **Natalie Foote** back to the team at the National Support Office. Natalie had been on maternity leave after having her second child and returned to her Executive Assistant role in July. It's great having her knowledge and experience back on the team, with her calm approach and can-do attitude.

Our Communications and Marketing Advisor **Melanie Louden** is leaving MDANZ after joining the National Support Office team in October 2020.

Melanie soon became our 'go to' girl at the National Office. Not only has she looked after our communications and marketing, she also took on extra duties in the Member Services and Executive Assistant areas while the roles were vacant.

We are so incredibly grateful for all her hard work and dedication. She has done an incredible job and we are going to miss her and all that she's brought to MDANZ.

A distraction and an escape

When Scott Boyle was dealing with pain, illness, death and PTSD, it was role-playing games that pulled him through and gave him a new world to immerse himself in.

So there I was – assembling a team of the most dangerous and efficient warriors in the galaxy.

I had a ship, a loyal crew, and the goal of stopping a mysterious race from abducting thousands of human colonists.

My crew and I fought countless battles and stared death in the face, laughing at its shadowy visage, and defying the odds by surviving an inconceivable suicide mission.

It was a journey of discovery and defiance.

And then, by some twisted form of cosmic humour, I was pulled from that world and back into ours by the wailing of sirens. Rushed to hospital in a blur of fatigue and pain, with that dark spectre snapping at my heels.

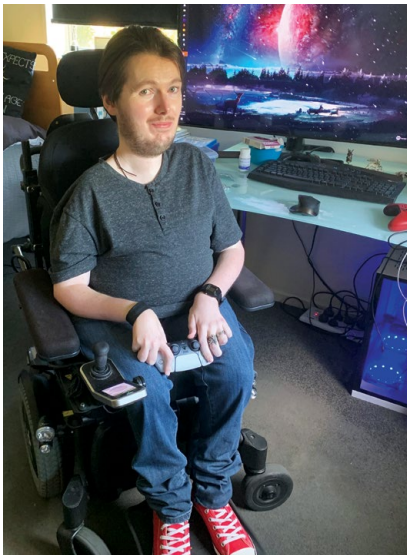
But this wasn't the first time, and it wouldn't be the last.

2011 was perhaps the most challenging year of my life. I suffered a horrible accident that left both of my legs and ankles fractured.

That trauma then led to multiple bouts of pneumonia, leaving me a tired and hollow shell of who I had once been.

In that year I almost died four times, and the effect of everything that happened cursed me with PTSD.

That part was genuinely worse than the accident and pain. In truth it took me years to manage and recover from the psychological torment.



Scott Boyle.

I made terrible choices to numb myself, pushed people away, and entered into bad relationships because I didn't care.

I had stared Death in the face, facing its formidable visage, but instead of laughing like in the game, I was broken.

So, why share this sob story?

Well, back then I had a lot of free time while my body became a battlefield.

I binged numerous shows, watched a lot of films, and burned through a small library worth of novels.

The pain from my legs was ever present and the chaos in my mind clouded my every thought.

I needed a distraction. An escape. And gaming delivered!

Until that year I had mostly played the likes of *Halo* and *Call of Duty*, focusing on multiplayer with my friends.

However now I needed something with a narrative, something that could help me forget the damage, if only for a few hours.

It was then that I discovered the beauty of role-playing games (RPGs), the genre that would dominate my future preferences and give me the power of choice.

In RPGs I could create my character and immerse myself in a new world, one where I wasn't a broken mess.

Those of us who endure our conditions have all dreamed about life without them and being capable of both ordinary and extraordinary feats. And thanks to those games, I was.

When I look back at that year and all the hardships I was subjected to, I realise that while my friends and family helped me immensely, gaming was what gave me an escape.

It helped me to cope with the pain and all the damage. And in the future, I expect it'll aid me again.

Scott Boyle is MDANZ's National Vice Chairperson.

A family favourite with a Mexican twist

Dietitian Julia Scott says corn fritters make a "super simple, tasty and healthy brunch, lunch or dinner".

Julia Scott is a dietitian with a passion for holistic health and encouraging people to live their best life.

"Eating well is important but so is getting our bodies moving, allowing ourselves to have rest, meditation and mindfulness, and fun with family and friends."

She believes the best diet is one you can stick to so her goal is to help people create healthy habits that lead to sustainable lifestyle changes.

Julia is a New Zealand Registered Dietitian. She has a Master of Science Majoring in Nutrition and Dietetics, and a Bachelor of Science Majoring in Human Nutrition. She is happy to share her recipes with MDANZ members.

Mexican Corn Fritters with Salsa and Avo

Ingredients

- ½ cup self-raising flour
- ¼ tsp salt
- 2 eggs, whisked with a fork
- 400g tin creamed corn

Corn is high in fibre, vitamin C, B vitamins, magnesium and potassium.

- 400g tin corn kernels, drained
- Pinch ground coriander
- 2 avocados

Salsa

- 1 tin chopped tomatoes
- ½ red onion, finely diced
- ½ cucumber, diced
- Juice of 1 lemon
- 2 Tbsp fresh coriander, chopped (or 2 tsp ground)
- Pinch of chilli flakes (optional)

Instructions

- Sift flour and salt into a mixing bowl.
- Add in beaten eggs and creamed style corn. Fold in whole corn kernels and coriander.
- Heat 1 Tbsp of olive oil in a frying pan over medium heat. Place spoonfuls of the corn fritter mixture into the pan and cook for 3-4 minutes until bubbles appear. Turn and cook the other side for another 3-4 minutes until golden and cooked through. Add more oil in between batches if needed. Keep warm in the oven.
- Mix salsa ingredients together in a bowl.
- Serve corn fritters topped with salsa and sliced avocado.

Makes 15 fritters.

You can follow Julia on
Facebook: @JuliaScottDietitian
Instagram: @juliascott_dietitian
Or on her website: www.juliascott.co.nz

Amazing opportunities for our youth members

There's a lot of hard work involved in completing the Duke of Edinburgh Award, but the effort is worth it, writes Award Leader Marty Price.

This last year has been somewhat different to the years since setting MDANZ up as a licensed award unit of the Duke of Edinburgh's Hillary Award in 2016.

Having Covid on our doorstep in 2020 and our country going into lockdown meant a whole new set-up for our Dukies who were unable to access their community and undertake their usual goals/sections of the award.

Our Dukies had to think outside the box and come up with ideas of doing goals from within their surroundings. Some were able to do this, whereas others had to wait to re-engage while the country went through the stages of getting back to Level One.

I commend all our Dukies that stuck it out and kept their motivation through this time as it was not easy for anyone, including myself as Award Leader.

This year more young MDANZ members are showing an interest in taking on the Duke of Edinburgh programme. We gained two new young Dukies earlier this year and I welcome them to this exciting opportunity that will open up new avenues for them.

The set-up for recording goals has improved over the years – all Dukies have an app which records the hours they do, and they can look over their progress in real time.

Marty Price.

I also have an app so I can oversee all the Dukies and their progress.

It gives me the opportunity to keep an eye on each member to see if they are working through things consistently; if not, I will get in touch to see if I can help them.

Sometimes other things like school exams, other activities or family matters might be taking them away from progressing with their goals. It's up to me to help where I can, to make it easier for our Dukies to get their goals signed off.

Currently five Dukies are doing the Bronze Award – Ryan O'Rourke from Gore, Joy Gutschlag from Nelson (you can read her story on page 20), Asher Hovell from Whangarei and Camille Peterson and Eden Hinchey, both from Auckland.

Two Dukies are doing their Silver Award – Dylan Schneider from Auckland and Jack Lovett-Hurst from Invercargill.

The range of goals our Dukies are taking part in include public speaking, photography, swimming, skim boarding, sailing, volunteering in a Sexuality And Gender Acceptance group, and cooking.

We will be working with them to look at what they would like to do for the Adventurous Journey – we can meet as a group over a weekend, or Dukies can complete it on their own over two days.

All Dukies have worked exceptionally hard and have done well to keep motivated. It is a lot of hard work, but the outcome is so worth it.

We are open to more MDANZ members taking part in the Duke of Edinburgh program, and Jack and Dylan would be great contacts for anyone wanting to chat about the amazing opportunities available through the award.

The Duke of Edinburgh's Hillary Award is open to MDANZ members aged 14 to 24. For more information go to: www.mda.org.nz and click on "What We Offer".

A cup of tea and a catch up with ... Jackie Stewart

Each issue we introduce a MDANZ team member.

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

I started with MDANZ in March 2020 – two weeks before we went into lockdown. I am the Fieldworker for the Southern Region, which covers Otago, Southland, Central Otago. I am here to provide support, information and encouragement to our members as well as educate the wider community about muscular dystrophy.

What qualifies as a great day at work for you?

Being able to achieve something that will, in some way, make a difference for our members. This can be an issue specific to an individual or something universal that may be affecting numerous members.

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

I would love to see our members enjoying the freedom and autonomy to make life choices according to their individual potential with all systemic and discriminatory barriers removed.

What are you passionate about?

Singing would be one of my greatest passions – I am currently a member of an acapella (unaccompanied) choir called Sunnyside Up. We sing mainly gospel and world music.

I also enjoy attending concerts and musical theatre.

I love plants and have lots of pot plants. I especially like the satisfaction of growing them from cuttings into large specimens.

During the months of daylight saving my husband and I volunteer in the community garden located within the grounds of our church. The fruit and vegetables are then put in a little kiosk for members of the community to access.

Finally, I love to walk although this requires a bit more effort and dedication in the winter months.

South Island wish coming true

Paige Subritzky is counting down to her big family holiday, writes the team from Make-A-Wish New Zealand.

For Paige Subritzky, a 13-year-old living with congenital muscular dystrophy, most of her life has been spent close to home with her family.

Through every treatment and surgery, her family comes together as her support system.

"We take every day as it comes. That's why we're so close as a family unit," says Paige's mother, Joycey.

The family stays close to home in Kaitaia and enjoys taking part in activities like fishing, diving and hunting.

When asked what her wish would be, Paige knew it would involve two things: expanding her bubble and including her family.

Make-A-Wish New Zealand is making the MDANZ Northern Region member's dream come true.

In the October school holidays, Paige and her family will fly to the South Island where they will begin their campervan adventure.

Paige has looked through maps, brochures and pictures with her siblings and started to make a list of activities they are excited to do.

"When I talk with my siblings about the places we want to go, that's when I get really excited about my wish."

Paige is looking forward to experiencing all kinds of new things: flying in a plane for the first time, seeing real snow, and even some

Paige Subritzky is excitedly planning her Make-A-Wish trip to the South Island with her family.

thrills like bungee jumping with her sister (if her mum will allow it, of course).

At Make-A-Wish, our mission is to grant the wishes of children who have critical illnesses to give them hope, strength and joy.

We believe that these wishes are transformational for the child and therefore all our work is based on positive psychology and through the implementation of what we call 'the wish journey'.

Every wish is unique and special, as is each child or teenager, and each wish is only limited by their imagination.

From going on scary rides with siblings or creating magical memories with family members, to princess playhouses, tree huts, bedroom makeovers, meeting celebrities or swimming with dolphins, the aim of Make-A-Wish is to always fulfil their one true wish.

For wish children like Paige, a wish not only creates an opportunity for them to have something to look forward to, but it also serves as a lasting positive impact for them and their families.

"This wish will be a lifelong memory for our family," says Joycey.

"We are really grateful for this wish and appreciate those that will help make this wish happen."

For more information about Make-A-Wish, or to apply for a wish go to www.makeawish.org.nz/wishes. If you would like to help Paige's wish to visit the South Island with her family come true, please donate today at www.makeawish.org.nz or by scanning the QR code below. Your donation will make an extraordinary difference for Paige.

Scan to help Paige's wish

DPO Coalition Update

Alison Riseborough, MDANZ's representative on the Disabled People's Organisation Coalition, provides members with an update on the latest issues.

Accessible voting – Electoral Commission: The DPO Coalition met with the Chief Electoral Officer to discuss ensuring that the 2023 General Election is fully accessible for disabled people.

Topics raised by the DPO Coalition included: Ensuring access to all information (including candidate information), accessible voting locations and online voting.

An election access fund is being created to support the participation of disabled people as candidates in Parliamentary elections. Consultation on the fund will be held this year and it will be open for applications in 2022. The Electoral Commission is updating its Disability Strategy 2020. The DPO Coalition looks forward to engagement as the new strategy is developed.

Reasonable Accommodations of persons with disabilities: The Reasonable Accommodations guide published by the Independent Monitoring Mechanism is being updated. The coalition has suggested that the revised guide could place more emphasis on the obligation of government and businesses to ensure equitable access.

It is also important for the guide to promote positive examples of reasonable accommodations being made. You can find the current guide here: www.ombudsman.parliament.nz/resources/reasonable-accommodation-persons-disabilities-new-zealand.

Learning Support Action Plan: The Ministry of Education is developing Terms of Reference for a review of learning support for students with the highest level of needs.

New roles called Learning Support Coordinators are being introduced. They are intended to build teacher capacity, provide support for students and are a point of contact for family/whanau. These coordinators work with schools' leadership teams to identify and plan for the needs of children who require additional support.

The Ministry of Education has also implemented a new practice model for learning support called He Pikorua which is available on the Ministry of Education website.

Literacy and numeracy strategies: The Ministry of Education is developing new strategies for literacy and numeracy. The goal of refreshing the strategies is to make the curriculum more bicultural and inclusive.

Health and Disability System Transition Unit work programme: In May the DPO Coalition began meeting with the Transition Unit responsible for implementing the health reforms. In June, the DPO Coalition and the Transition Unit discussed the draft work programme. This identifies target areas, immediate opportunities and actions. The DPO Coalition will continue to meet regularly with the Transition Unit.

Accessibility legislation: The coalition also continued its regular engagement with the Ministry of Social Development policy team about options for accessibility legislation. This work has the potential to set down the Government's requirements for what all parts of New Zealand society, business and government must do to deliver accessibility for disabled people on an equal basis with others. The coalition learned the Minister for Disability Issues updated her Cabinet colleagues in May, and they are broadly supportive of the approach towards accessibility legislation.

Opt-in guidelines to support disabled people to attend arts, culture and sporting events: The Ministry of Social Development would like to better understand the needs of disabled people attending events, and the needs of event providers. It intends to develop a plan that will enable disabled people to take a "companion" to events and performances at a lower or no cost.

You can contact Alison at alison.riseborough@mda.nz

Ready, set, roll

Neil Singh is ready to roll for our annual appeal

Roll 1k for MDA

20-26 September
www.roll1kforMDA.org.nz

This year's national annual appeal is something wheelie fun, interactive, and even better – anyone and everyone can take part, writes *Melanie Louden*.

It relies on you

Roll 1k for MDA is a fundraising and awareness campaign that relies on you, our valued *In Touch* readers, rolling 1km on any kind of wheels and fundraising amongst family and friends.

Simply head over to **www.roll1kforMDA.org.nz** to register and create your own fundraising page. Then spread the word via email and social media, asking people to either sign up to fundraise themselves, or donate to your fundraising page.

You could roll 1km on a wheelchair, electric scooter, skateboard, roller skates, push bike … or whatever creative wheel idea you can come up with.

Spend the month of September gathering donations – and once you've raised $50 you'll receive a free **Roll 1k for MDA** t-shirt. Complete your roll during the week of September 20-26.

All the funds raised will go to the region they were collected in to contribute to our vital Fieldwork Service which provides essential support to members and their families.

We're delighted to have sponsors providing wheelie awesome prizes for the individual and team that raises the most money.

Northern Region member Neil Singh is the face of the campaign and his parents mum Yasha Aggarwal and dad Kuldeep Singh are sharing their story to help raise awareness and funds.

Neil's story: When you meet Neil Singh for the first time you can't help but be blown away by this young man.

He's 10 years old, yet he has the confidence of a wise and inquisitive person.

He's only 109.5cm tall, yet he commands the room with his charming, extrovert personality. Neil loves to ask questions and can hold a meaningful conversation with any one of any age.

He has a love of trains, music and Lego, Pokemon and Beyblades.

He also has Duchenne muscular dystrophy - a progressive and genetic muscle disease that affects approximately one out of every 3600 baby boys.

It affects many parts of the body, which results in deterioration of the skeletal, heart and lung muscles.

When Neil wasn't reaching his toddler milestones, mum Yasha Aggarwal and dad Kuldeep Singh knew something wasn't right.

"He was late in standing, walking, running from the get-go, but every time we were reassured by health professionals that he was doing things in a normal time frame, and we did not need to be worried about his development," Yasha says.

They asked again to see a Starship doctor and this time they happened to meet with a pediatrician who was familiar with Duchenne, although Yasha and Kuldeep didn't know that at the time.

Days later, when it was time to go back to Starship to get the results of the tests, Yasha took Neil to the appointment by herself only to realise she was going to be seen by the neuromuscular clinic instead of general pediatrics.

"Things did not feel right, and my concern grew while I waited. It was at its peak when they said that Neil can go to the playroom while I discuss the results with the doctors, who then told me the diagnosis. It was Duchenne.

"I was without Kuldeep listening to the diagnosis. I was not told to bring a support person.

"I felt sad, angry and was in disbelief. They told us that our beautiful boy had a terrible condition."

Yasha was given some space to take in the news and call her husband, who rushed to Starship to be with his family.

"We sat, holding each other, crying and trying to pull ourselves together before we saw Neil again," she says.

In need of support, Yasha and Kuldeep reached out to the Muscular Dystrophy Association of New Zealand where they met other parents who knew what they were going through.

"They knew everything, every emotion without us mentioning it," the couple say.

"MDANZ has held our hand and helped us with various stages of Neil's condition, getting information. The Fieldworkers have been available as a support person when we needed them."

Neil now has a power wheelchair that he uses at school and for long distances. He gets tired easily and his parents must watch his activities throughout the day so they can manage his fatigue.

"He would love to play soccer, or just run around, and play video games with his buddies. Unfortunately, regular soccer is not an option. This is due to his reduced ability to run like other children, fatigue and, most importantly, safety.

"His hands get tired if he plays video games, his legs get tired if he runs around."

'Fatigue management' played a big part in Neil starting school and while Neil was very excited about it, his parents were quite anxious.

However, the move to school life couldn't have gone better with the school being very engaged and approachable, and doing an excellent job of communicating with Yasha and Kuldeep, which helped everyone settle in.

Five years down the track and Neil "loves" school, with art and music being his favourite activities.

"He has a close group of friends that he looks forward to meeting every day. He is performing well and reaching his goals."

Yasha and Kuldeep say Neil is aware that his condition will progress. "He talks about not being able to walk one day, which is heartbreaking for him and us."

However, the family is staying strong and united. "We are taking each day as it comes, and we prefer not to look too far ahead in the future.

"The prognosis is not hidden but we'd rather not think about it."

Photos by Hamish Melville.

Jezza Williams, front, is a tetraplegic who is an expert in inclusive tourism.

Yes, you can… raft, paraglide, sea-kayak

Taking inclusive travel to the next level

From an able-bodied expert in adventure tourism to a world expert in inclusive tourism and travel, Jezza Williams shares his love for ensuring everyBODY can experience adventure.

Recently I was talking with a young woman about why she doesn't get out and enjoy adventure.

Her reply was: "Being born with a disability, everything was impossible when I was a child, so it was put in the too hard basket and that's where it stayed."

Well, things have changed, and adventure experiences are now becoming inclusive to all.

I feel privileged. Ever since my feet touched the ground they wanted to adventure.

In fact, following my passion took me on a journey of two decades of guiding internationally on spectacular rivers and canyons, adventuring into lost jungles, patrolling steep and deep mountains.

Working in wild environments, I chose a life of risk-taking and exploring limits.

Adventure has taught me life lessons, it prepared me for anything and encouraged me to live to my potential.

So, when a life-changing canyoning accident in the Swiss Alps left me with a C5 spinal cord injury, I was ready to accept the challenge.

After a lifetime of experience in the adventure industry my initial fear was that I'd never be able to get amongst the action again.

After researching the possibilities for someone that was in my new predicament to experience adventure, I was blown away at the lack of opportunity and infrastructure.

As a tetraplegic, with my experience I'm the perfect guinea pig, realising what is possible, practical and safe. So, if my body can do it, most bodies can do it.

Changing my focus, I decided to open the adventure industry to all and in 2012 Makingtrax was born.

I originally opened with the experiences that I knew, and funnily enough these were the most difficult – rafting, paragliding, sea kayaking etc.

Makingtrax focuses on the experience rather than the infrastructure.

It is called Inclusive Tourism, a little different than Accessible Tourism where the focus is on infrastructure. In the adventure industry making everything accessible is just not possible or practical.

Inclusive Tourism is about opening possibilities. We do this through:

- Education – to the industry.
- Information – to you the client.
- Cooperation – between the operator and you.
- Adaptation – only if required.
- Promotion – providing awareness to possibilities.

As a tetraplegic, with my experience I'm the perfect guinea pig, realising what is possible, practical and safe. So, if my body can do it, most bodies can do it.

The Inclusive Directory on the Makingtrax website has all our Inclusive Movement Operators in one place and relevant information for anyBODY to have the confidence to book their experience.

We believe everyone should be able to rock up to our inclusive operators and enjoy an adventure. All adaptive equipment is located at the inclusive operators.

Makingtrax also designs and manufacturers support seats and other adaptive systems to open up everything from whitewater rafting through to paragliding.

A little taste of the most popular destinations:

- Queenstown: Paragliding to packrafting, canyon swinging or the world's only commercial jet sprint are just a few.
- Franz Josef: Snow landings with a ski chair, skydiving 20,000ft above, or sea-kayaking through ancient rainforests.
- Abel Tasman: Golden beaches, sea kayaking eco tours or a waka cultural experience.
- Auckland: Whitewater park, whale encounters or skydive from 20,000ft.

There are no excuses, so get busy living!!

Makingtrax recently became a charitable trust – The Makingtrax Foundation. Our vision is to see all adventure operators, outdoor educators, the tourism industry, Department of Conservation and cycleways embracing inclusion, whilst having adoption and education in place to make these experiences possible for everyBODY. To learn more about Makingtrax go to www.makingtrax.co.nz.

Makingtrax has opened up all types of outdoor adventure activities for people with disabilities.

Life-changing throat surgery a turning point

Eating difficulties, combined with mobility issues, were having a major impact

A diet of soft pureed food has been replaced with an almost normal diet, writes MDANZ Canterbury member Mary Fargher.

My name is Mary Fargher, and I am 74 years old. I was diagnosed with inclusion body myositis (IBM) in 2016 following an MRI, nerve conduction tests, and then a muscle biopsy taken from my upper arm.

Inclusion body myositis is an inflammatory muscle disease characterised by chronic inflammation with slowly progressive weakness of both the distal and proximal muscles.

It is most apparent in the muscles of the wrist, fingers and thighs.

Prior to my diagnosis I had experienced weakening of my arms and thighs for a period of three to four years and was prone to falling, tripping and it was increasingly difficult to get up off chairs or the floor.

I had been attending a pilates class at a local physiotherapy clinic, and although the instructor (a physiotherapist) gave me extra exercises to do, it did not help. She was concerned enough to consult with my GP who then referred me to Dr Desiree Fernandez, a consultant neurologist.

I am now unable to walk very far without a walking stick and have a mobility scooter. I purchased one that can be dismantled to fit into the boot of a car which is great for going further afield than just around the block.

My left hand and arm are very weak, and I am unable to carry anything of any weight. Having been an active gardener and quilter in the past this has impacted my life considerably.

Mary Fargher can eat and drink with confidence following surgery.

Over the last five years I have also developed dysphagia (difficulty swallowing).

Swallowing became increasingly difficult with constant choking and coughing on solid food as well as drinks.

This resulted in me having to cut back on socialising and eating out and having to resort to a diet of soft puréed food.

Following a referral from a Nelson ENT surgeon to Mr Robert Allison at Christchurch Hospital, I underwent a pharyngoscopy and cricopharygeal myotomy in November 2020.

My social life has returned, and I can eat out and enjoy a good coffee again without continually worrying about choking on anything and everything.

This operation involves the surgical sectioning of the upper esophageal sphincter. Dysphagia stops the esophageal sphincter from relaxing to allow food to enter the esophagus or it relaxes in an uncoordinated manner.

The difficulties with eating combined with my mobility were having a major impact on my life.

The throat surgery was a huge turning point for me and within two to three weeks I was able to return to an almost normal diet again.

I have a very unobtrusive scar in the folds of my neck which does not cause me any issues.

I was in hospital for two nights and although a bit uncomfortable for a few days it was nothing compared to the life-changing effect it has had for me.

My social life has returned, and I can eat out and enjoy a good coffee again without continually worrying about choking on anything and everything.

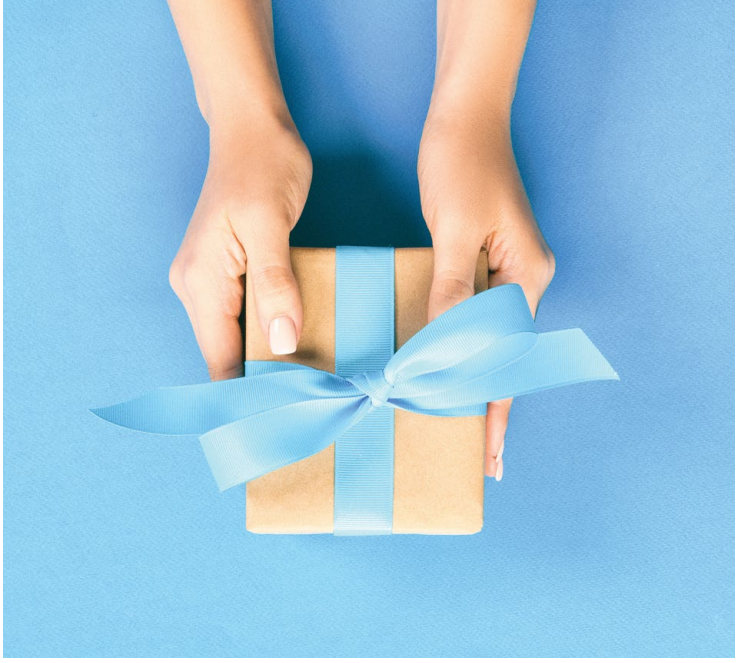
The surgery didn't create very much anxiety for me as I was desperate to try anything which might help.

Due to the Covid-19 lockdown I had to wait 10 months for the surgery so I had a lot of time to think about it.

A biopsy taken from the muscle in my throat confirmed that it was the IBM that had caused the eating difficulties that I had been experiencing.

If I have these issues again, I have been advised that there is every likelihood that I could have a repeat procedure.

I became a member of MDANZ in 2016, just after my IBM diagnosis and have found the Fieldworkers, the *In Touch* magazine, and regular emails, particularly in regard to Covid issues, informative and reassuring.



Young, strong and living independently

Brittney is giving back to the community that supported her

Brittney Steele has a simple plan when it comes to succeeding in life – just "keep trying". *By Melanie Louden.*

Brittney Steele has always been determined, persistent, and wanting to live life her way.

So, it's no surprise that the 24-year-old Invercargill resident lives on her own and works in a job that sees her giving back to the community that helped her, while living with Friedreich ataxia.

Brittney was diagnosed with the condition on New Year's Eve, when she was seven-years-old.

"Telling a seven-year-old she would have a wheelchair in the next couple of years didn't sound horrible, because I didn't fully understand," she says.

"Mum and Dad always knew there was something a little different with me. I was 'lazy' (my words not theirs) - I would rather stay in and watch TV than go outside and run and play with my two older sisters.

"When I would go to the toilet when I was younger [before diagnosis], I would come back to what I was doing with no knickers on. We didn't realise till years later that it was because I couldn't hold my balance on one leg to put them back on."

Brittney, who grew up in Mataura, Southland, got her first manual wheelchair when she was 10, and an electric wheelchair the following year while living with her dad in Cromwell.

"At first I thought it was awesome because I didn't have to be in it all the time," she says.

Brittney later returned to Mataura and recalls her two older sisters, Jamie and Shannon, being very protective of her when they were all at college together.

"They carried me on and off the school bus – my chair stayed at school and I used a walking frame at home or my manual chair, depending on how tired I was."

Brittney Steele is "obsessed" with candles so rather than spending money on them, she started making them. Photo: Shannon Steele.

They were also quick to step in if anyone was mean to Brittney or "looked at me funny". She says it was a comfort to have them there, but at the same time she "hated school".

Brittney initially left school when she was 16, but went back six months later and stayed till the end of year 13, at which point she moved to Invercargill.

"Ever since I was little, I didn't want mum to be my caregiver and I didn't want my family to look after me."

So, she moved into accommodation provided by Pact.

Pact's website says it provides "support to anyone with a need so they can lead fulfilling lives in the community" – everything from family/whanau support, respite, addiction support through to accommodation and community support, and much more.

"I moved to Pact because it was the only place I could go. I moved in and out a few times."

Brittney last moved out of Pact more than three years ago and since then she has been living in a two-bedroom flat with her cat Craig (named, simply, because it wasn't a common name for a cat).

She was determined to work at Pact, keen to give back to the organisation that helped her, and she now works there four days a week as the youth consumer advisor.

"When I lived there, I always wanted to be staff there. So they made a role just for me. I'll go to the youth home and hang out, I'll spend a day in the office, I'll organise things for the small groups."

And the job is rewarding. "Oh, I love it. It's awesome. When I was at the youth home, I would have wanted someone [that worked there] who was young and knows what it's like."

When she's not working, Brittney can be found making her own candles. "I'm obsessed with them. I used to spend so much money on them so I started making them two years ago."

She used to sell the candles on Facebook, but when that got too demanding she switched to making them for pleasure and giving them away as gifts.

"I love it because this is something I can do. It's super easy."

Brittney can make candles with any scent requested and says her favourite are the ones that smell like sweets – especially Hubba Bubba and white chocolate.

"I love looking in second-hand shops for little tea cups and saucers, and jars [to make candles in]. I like it because it's something I made, which makes it even cooler.

"I can't really cook anything, but I can make candles."

Brittney says she's proud of the fact that she does a lot of things that young people her age normally do – like living by herself and making her own money.

She says the key to making the most of life is being persistent. "If I want something done, I have to keep trying. I just have to do it. Even if it's taking for ever."

Research Grants 2021

Neuromuscular Research New Zealand

is once again accepting funding applications for research relevant to New Zealanders living with neuromuscular conditions.

Proposals most likely to be considered will be those which address standards of care (including best and/or innovative practice in supporting people with neuromuscular conditions); effectiveness and/or cost benefit of service delivery modes and interventions; prevalence and incidence of neuromuscular conditions; and proposals which extend existing research into treatment and cures.

Research of a preliminary nature with the intention of developing further proposals for substantial financial support from elsewhere will also be favoured.

Closing date for applications is 17th September each year and we hope to advise the outcome by 1st December.

Register your application in advance by emailing nrft@mda.org.nz or go to the MDA's website www.mda.org.nz/Our-Research/Apply-for-Funding

Neuromuscular Research New Zealand

Creative and practical

Finding solutions with a 3D printer

Funding from the Bradley Jenkin Memorial Fund means Daniel Lyall can keep making 3D items that make life easier for himself, and for other MDANZ members.

Switching from viticulture work on a vineyard to creating things with a 3D printer might seem like an unusual leap, but for Daniel Lyall it's all about independence and quality of life.

The Blenheim husband and father says it's about making life easier and making practical items cheaper.

3D printing, or additive manufacturing, is the construction of a three-dimensional object, from a CAD (Computer-Aided Design) model or a digital 3D model, out of plastic, one layer at a time.

It can be used to make all sorts of items such as furniture, wax castings for making jewellery, tools, tripods, gift and novelty items, toys – and wheelchair accessories.

Daniel, who has Becker muscular dystrophy (BMD), has made fun things – like a miniature Tardis light – as well as practical things like a drink holder and a side table for his wheelchair.

He says making things isn't easy – he's had plenty of failed attempts, and he went on a course to learn how to manufacture things properly.

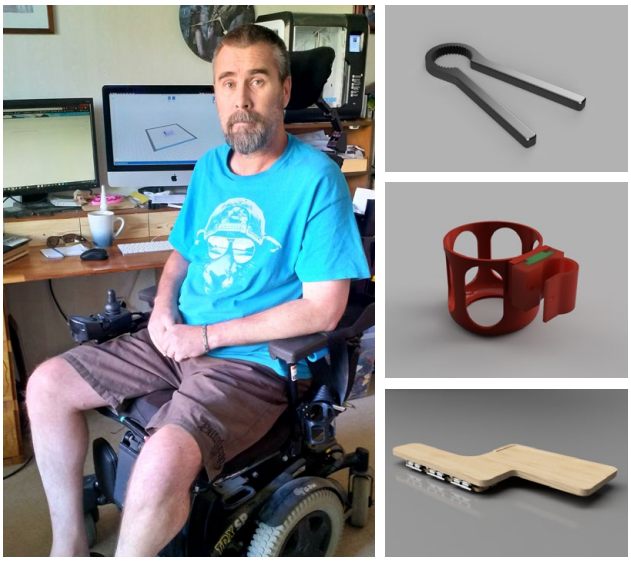
"You've got to know how to design them in the first place," the 43-year-old says.

The drink holder can be made in a day and uses brackets that mount to a tube on the arm of a wheelchair. He's made cup holders of different sizes for different cups.

Daniel Lyall's minature *Dr Who* Tardis light.

The side table folds down beside the wheelchair when it's not in use so it doesn't take up any extra space. Just like the cup holder, the whole thing, including a three-piece hinge, have all been made on his 3D printer. Daniel also has plans to make a mechanical side table attachment that will move into position and away again with the push of a button.

He says both items are easy to use as well as practical and, depending on a person's level of mobility, they won't



Daniel Lyall at work. He has manufactured (from top to bottom) a bottle opener; cup holder and side table.

Daniel says 3D printing means things can be made at a fraction of the cost of purchasing them commercially, and he's keen to sell his items to MDANZ members.

need a carer to move or remove each item.

"It's all about independence and quality of life," he adds.

Daniel, who worked on vineyards from the age of 18 to 32, was diagnosed with BMD when he was 28 years old. The diagnosis came after he gathered with family for Christmas and he learnt a cousin had been diagnosed with BMD.

"I knew something was wrong with me. I couldn't ride a bicycle anymore. I thought [my cousin's diagnosis] might explain things.

"But it took a long time to come to terms with it."

When Daniel spoke to *In Touch* he had a few projects on the go – a tablet mount, a wheelchair attachment that could help people move their arm, and a robot arm.

Daniel uses a design programme called Fusion 360 and is considered one of their expert users (Autodesk Expert Elite).

He helps users make, fix and find solutions to whatever it is they want to make.

His 3D inspiration came about after he was talking to then Fieldworker Paul Graham about how expensive things like wheelchair accessories were.

"I looked at them and thought I could make that much cheaper. I couldn't get what I wanted – so I made it. It all started from what I really needed at the time."

Daniel, who has always enjoyed woodwork as a hobby, received funding from MDANZ's Bradley Jenkin Memorial Fund in 2019 to buy the 3D printer he currently uses.

"I asked for funding because the printer I had at the time wasn't all that great. I had lots of fails with it and it wasn't user friendly. The new one is easier to use, it's enclosed and it makes complex shapes easier to make. It just makes things better. It can use fancier carbon fibre impregnated filaments which makes stuff really strong."

He says 3D printing means things can be made at a fraction of the cost of purchasing them commercially, and he's keen to sell his items to MDANZ members.

Daniel has been a member of MDANZ for 15 years now and is grateful for the support of the Fieldworkers.

"They've have given me ideas on how to make things easier and what to do - they have suggestions. They are doing a job they want to do."

He appreciates "knowing someone is there that I can talk to" and that they can help get things sorted when issues arise.

Anyone who wants to discuss buying 3D printed cup holders and side tables from Daniel can contact him via email - daniel_lyall@hotmail.com or check his website www.danielswheelchaircustomisations.weebly.com/

The wheelchair side table and cup holder.

The Bradley Jenkin Memorial Fund

The Bradley Jenkin Memorial Fund helps MDANZ members with a neuromuscular condition receive funding for access opportunities and specialised resources that enable them to achieve freedom.

The fund has helped members purchase specialised sports equipment, participate in sporting events, it has contributed towards the cost of obtaining a mobility dog, provided mobility equipment, and assisted with career development such as university and course fees.

For criteria, info and to apply, go to **www.mda.org.nz** > What We Offer

Funds must be spent within three months.

Applications close

January 31 • April 30 • July 31 • October 31

Joy's journey to leadership

Real life lessons learnt on Duke of Ed programme

Joy Gutschlag has made the most of the many opportunities offered by the Duke of Edinburgh's Hillary Award.

Taking part in the Duke of Edinburgh's Hillary Award has produced plenty of positive spinoffs for Joy Gutschlag.

She's moved out of home and therefore has had to take responsibility for managing her condition, she's enjoyed having a positive impact on the people around her and, the usually reserved 20-year-old, has had to "step out".

She has also been able to indulge in her love of music.

In 2018 Joy was diagnosed with muscular dystrophy, although she hasn't yet received a full diagnosis, and she promptly joined MDANZ.

When the Nelson resident became a member her then Fieldworker Marty Price came to visit. Marty is also the organisation's Award Leader for the Duke of Edinburgh programme and encouraged Joy to get involved.

"I liked the idea of being able to partake in it and do it while having a disability. I think it's really cool that you can do something like this, even with a disability. I got to do things I otherwise wouldn't have done," she says.

"For me the biggest thing is that I got to take part in something. I felt included."

The Duke of Edinburgh's Hillary Award gives 14 to 24-year-olds the chance to challenge themselves, learn new skills and meet new people.

Participants carry out tasks in three categories – physical recreation; service; and skills, and go on an Adventurous Journey, to complete bronze, silver, and gold levels.

They design their own programme, set goals and record progress towards their achievement.

Joy has completed the bronze section by choosing swimming for her physical recreation activity, being a youth group leader for Unite church for her service activity, and attending the Discipleship Training School at Youth With A Mission (YWAM) for her skill.

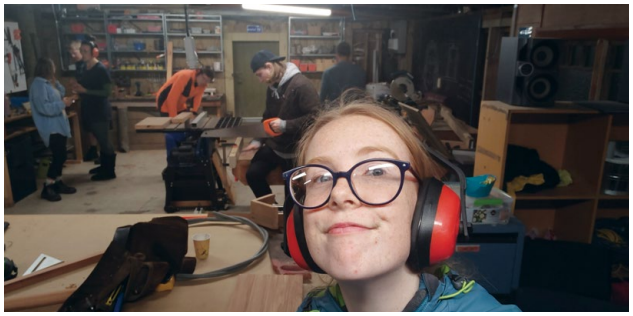
Joy Gutschlag is making the most of "stepping out" thanks to the support of the Duke of Edinburgh programme.

She says the swimming kept her focused and committed to a routine, while the service aspect "pushed me out of my comfort zone as a leader".

"I had a bunch of teenage boys. I had no idea how to do it. I'm more of a reserved person and I really had to step out. I had to take ownership and responsibility."

Completing the Discipleship Training School for the Duke of Edinburgh skill activity meant moving out of home in October 2020, at the age of 19, and living at the YWAM campus.

That was a big step for Joy, who had been home-schooled, as she had to take greater ownership of managing her condition.



Joy Gutschlag has been supporting and encouraging teenagers as they build tiny homes for Nelson's homeless.

In the process, she's also learning an important lesson. "It's okay to not keep up."

Not only does Joy study and live at YWAM (which equips people to become missionaries) she also works there as a volunteer.

She is currently supporting and encouraging teenagers as they build tiny homes for Nelson's homeless.

"It's cool to know you're having an impact."

Joy says she is looking forward to the Adventurous Journey section of the award, where the MDANZ Duke of Edinburgh participants get together for an adventure, particularly because she is yet to meet any other young people with muscular dystrophy.

Joy has developed an interest in music over the last three years, she's learnt to play the piano and the guitar, and has since picked up the violin.

Being a youth leader meant she had more opportunity to play, and "learn the skills".

She used to lead the youth group band and jokes, "I was very…not good".

"Even though I'm terrible, they let me have a go. Now they don't block their ears. It's been really cool to have that opportunity."

Joy encourages other 14 to 24-year-olds to consider taking part in the Duke of Edinburgh programme.

"Get out there and do it. Do things that you enjoy doing – it's a cool opportunity to do that."

Joy's health issues started when she was still very young.

"When I was 7-10 years old, I had a kidney condition that made me really sick. My growth had been stunted because I had been so sick."

Then when she was 12-13, Joy grew really fast and her knees turned in. She had been keen on figure skating, but couldn't keep that up any more.

When she was 18 years old, she was diagnosed with muscular dystrophy.

She says living with the impact muscular dystrophy has on her everyday life is "just normal for me", however, it does mean she gets a lot of pain.

"I wear AFO's (ankle foot orthosis) and I don't have much muscle, so I have to watch every step I take so I don't fall."

Joy is grateful for the support she gets from MDANZ and says meeting Marty and her current Fieldworker, Jane Hazlett, has been very important.

"When I was diagnosed I knew nothing, I didn't know what muscular dystrophy was. It was really comforting to have someone there."

MDANZ is a licensed Hillary Award provider, and can provide funding and direct support for members aged between 14 and 24 years who want to take on the challenge to achieve either bronze, silver or gold level awards. For more information go to https://www.mda.org.nz/What-We-Offer/Duke-of-Edinburghs-Hillary-Award.

The Duke of Edinburgh programme has helped Joy Gutschlag indulge in her love of music.

Finding help entering the workforce

The Be. Lab team explains how they are on hand to help people with access needs, or long-term health conditions, into employment.

Be. Lab has a vision for New Zealand to become the most accessible nation in the world and central to that vision is connecting talented access citizens with outstanding employment opportunities.

Access citizens are a massive, untapped pool of talent who have the skills that workplaces in the 21st Century need. This outstanding group has an enormous diversity in areas of interest, tertiary education and work experience.

In partnership with the Ministry of Social Development, we offer several unique employment programmes for those with access needs or long-term health conditions.

Much more than a recruitment service, our programmes offer tailored wrap-around support including professional development, access to an external mentor and ongoing personal support to ensure success.

Our Industry Partnership programme supports access citizens in their quest to find full-time work with a particular focus on the tech, professional services and health sectors.

Our Internship Programme places access citizens in a paid 12-week internship in the career of their choice. Available to recent graduates or those in their final year of study, 73 percent have gone on to secure permanent employment.

MDANZ Northern Region member Craig Pollok works for Ricoh NZ.

To those with access needs, please know there is a different purpose for everyone. We all have our own paths but with the right support we can have a great career.

We are also thrilled to extend this highly successful Internship Programme to a wider range of access citizens – those who don't have a tertiary qualification and are seeking work experience.

Craig Pollok who lives with MELAS, a form of muscular dystrophy, was supported by Be. Lab in 2016 and landed a job with Ricoh NZ.

Craig explains that MELAS has caused him to have five strokes over his life so far which affects his ability to walk, his balance and his speech by weakening the connection between the brain and other parts of the body. The impacted speech has affected his work life as people have to make an extra effort to understand him.

Here's what he has to say:

"Be.Lab gave me the opportunity to kick-start my career and helped make my transition from university to working life as easy as possible.

"Be. Lab and Ricoh NZ made it easy for me to adjust to the working world by providing assistance such as acquiring a standing desk.

"I'm loving my job and have created a personal development plan with my manager to develop goals that help me be the best I can be.

"To those with access needs, please know there is a different purpose for everyone. We all have our own paths but with the right support we can have a great career."

If you are an access citizen or have a long-term health condition and need support with your career, get in touch with Be. Lab today. Email talent@belab.co.nz or phone 09 309 8966. For more information visit www.belab.co.nz.

Work and Income: What it can do to help you

Work and Income helps tens of thousands of New Zealanders who need support with a health condition, illness, or disability – including people with muscular dystrophy. Here, Work and Income explains some of the ways it can help MDANZ members.

Work and Income is here to help, and does so every day for people facing hardship because of a health condition or medical costs.

Depending on your circumstances, you may be eligible for financial assistance. In addition to a benefit, you may also be eligible for other supplementary assistance to help meet living costs or other costs related to having muscular dystrophy.

This assistance does require Work and Income knowing your income and assets to determine what you may be eligible for and, in some instances, what you may need to pay back.

Some of the assistance available:

- Disability Allowance – a weekly payment for people who have regular, on-going costs because of a disability, such as visits to the doctor or hospital, medicines, extra clothing, or travel.
- Child Disability Allowance – a fortnightly payment to the main carer of a child or young person with a serious disability, in recognition of the extra care and attention needed for that child.
- Childcare Subsidy – may be paid up to 50 hours a week if the principal caregiver has a condition which results in reduction of their independent function and that condition is expected to continue for at least six months and they do not have a partner who is able to provide childcare or there is good and sufficient reason why the partner cannot provide childcare, plus extra provisions for people with a special needs child or sibling in hospital.
- OSCAR Subsidy – may be paid for up to 20 hours a week in the school term or 50 hours a week in the school holidays if the principal caregiver has a condition which results in reduction of their independent function and that condition is expected to continue for at least six months and that illness or disability is so serious as to warrant additional childcare, plus extra provisions for people with a special needs child (max age increases).
- Residential Support Subsidy – helps with the cost of residential support for a person with a physical, sensory, intellectual, psychiatric disability or disabling chronic health condition who needs residential care as a result. The subsidy is paid to the residential service provider by the Ministry of Health to help with the cost of residential care.
- Accommodation Supplement – a weekly payment helping people with their rent, board or the cost of owning a home.
- Temporary Additional Support – a weekly payment helping someone who can't meet their essential living costs from what they earn or from other sources.
- Special Needs Grants – non-taxable, one-off recoverable or non-recoverable financial assistance to meet immediate needs. A client does not have to be receiving a benefit to qualify for Special Needs Grants.
- May be eligible for Family Tax Credits.
- Community Services Card helps with the costs of some health services. It is granted automatically to people getting a benefit. People on a low income can also apply for it.
- Residential Care Subsidy, which is paid through the District Health Boards by the Ministry of Health, assists with the cost of contracted care for a client needing age-related long-term residential care in a hospital or rest home indefinitely. Generally, the amount of subsidy is the difference between the cost of contracted care and the amount a client is required to contribute for that care.

You can find out more through Work and Income's online eligibility tool: www.workandincome.govt.nz/online-services/eligibility/.
If you're in need of assistance, please contact Work and Income on 0800 559 009 to talk about your individual situation.

Your MDANZ Fieldworker can assist you with approaching Work and Income for support.

Multidisciplinary approach to managing EDMD

Emery-Dreifuss muscular dystrophy affects both men and women, with first symptoms usually appearing between the ages of 17 and 40.

Emery-Dreifuss muscular dystrophy (EDMD) is a condition that mainly affects muscles used for movement (skeletal muscles) and heart (cardiac) muscle. It is named after Alan Eglin H. Emery and Fritz E. Dreifuss.

Among the earliest features of this disorder are joint deformities called contractures, which restrict the movement of certain joints. Contractures can become noticeable in early childhood most often involving the elbows, ankles and neck.

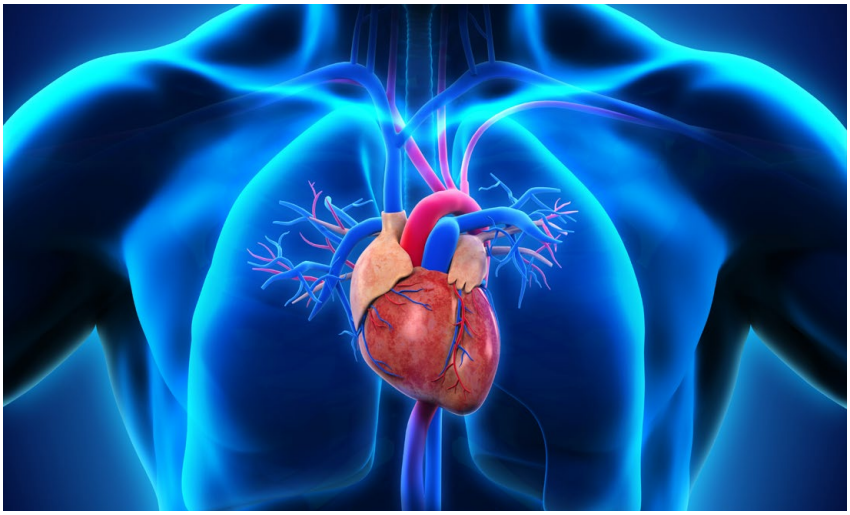
Most affected individuals also experience slowly progressive muscle weakness and wasting, beginning in the muscles of the upper arms and lower legs and progressing to muscles in the shoulders and hips.

Almost all people with EDMD have heart problems by adulthood, heart conduction defects and abnormal heart rhythms. Untreated, these abnormalities can lead to an unusually slow heartbeat (bradycardia), fainting (syncope), and an increased risk of stroke. Occasionally, sudden cardiac arrest is the first symptom of the condition.

This disorder affects both sexes with first symptoms usually displayed between the ages of 17 and 40.

Types of EDMD

There are three types of EDMD



distinguished by their pattern of inheritance: X-linked, autosomal dominant, or autosomal recessive.

X-linked: X-Linked EDMD is caused by a defect in either the FHL1 or EMD gene on the X chromosome. As boys have only one X chromosome if they inherit an X chromosome with the defective gene then they will have X-Linked EDMD.

The mother is described as a carrier and with one functioning EMD gene is usually unaffected, although can have the related heart problems. X-Linked EDMD is the most common form of this condition, affecting an estimated one in 100,000 people.

Autosomal dominant: The LMNA gene is located on chromosome 1 and is considered to have an autosomal dominant pattern of inheritance i.e. one defective copy is enough for the disease to present. Men and women are equally affected. Some 75 percent of this form are caused by new mutations.

In the remaining cases, people inherit the altered gene from an affected parent. Autosomal dominant EDMD is thought to be more variable than the other types, with a small percentage of people with this form experiencing heart problems without any muscle weakness or wasting. The incidence of this form is unknown.

Autosomal recessive: Rarely, LMNA gene mutations can cause a form of EDMD that is inherited in an autosomal recessive pattern. This means that both copies of the LMNA gene must be defective for

the disease to develop fully. In this situation each parent is a carrier of one defective gene. This form of EDMD appears to be very rare with only a few cases reported worldwide.

Causes of EDMD

Mutations in three different genes (EMD, FHL1 and LMNA genes) cause EDMD.

The EMD gene codes for a protein called Emerin, a serine-rich nuclear membrane protein essential for the normal function of skeletal and cardiac muscle. Most EMD gene mutations prevent the production of any functional Emerin. It remains unclear how a lack of this protein results in the signs and symptoms of EDMD.

The LMNA gene codes for two proteins - Lamin A and Lamin C. They are found in the cell's nucleus and provide structural and regulatory function in the nuclear membrane as well as regulation of certain genes in the cell nucleus. Most of the LMNA mutations that cause this condition result in the production of an altered version of these proteins and researchers are investigating how they lead to muscle wasting and heart problems.

The FHL1 gene codes for a protein called Four and a half LIM domains protein 1. This protein is heavily produced in skeletal and muscle tissue.

Most cases of EDMD are caused by mutations in the EMD gene. Less commonly, EDMD results from mutations in the LMNA gene.

Genetic Counselling

Genetic counselling is available to families who have had a diagnosis of EDMD. As there are several different inheritance patterns it is important that the diagnosis is correct. You can access this via your GP, self-refer or an MDANZ Fieldworker.

Diagnosis

You will need to see your doctor who will try to determine if the weakness is a problem with the muscles or the nerves that control them. Other tests may be ordered, and you will most likely be referred to a neurologist for review. More specialised tests may be undertaken. These may include:

- Nerve conduction studies and electromyography (EMG). In these tests, electricity and very fine pins are used to stimulate and assess the muscles or nerves individually to see where the problem lies.
- Muscle biopsy (surgical removal of a small sample of muscle from the patient). Modern techniques can be used to distinguish muscular dystrophies from infections, inflammatory disorders and other problems.
- Molecular genetic (DNA) testing of EMD, FHL1, and LMNA.

Management

There is a multidisciplinary team approach to the management of the patient and their symptoms during the course of this condition. A neurologist, cardiologist, respiratory physician, physiotherapist, occupation therapist, orthopaedic surgeon and a dietitian may all be needed at some point. Timely referrals are important. Interventions include:

- Physical therapy and stretching to prevent contractures.
- Maintenance of an appropriate weight.
- Medical intervention to prevent cardiac complications.
- Surgery may be required to release contractures and to manage scoliosis (curvature of the spine).
- Aids to assist in walking e.g. canes, walkers, orthoses, wheelchairs.
- Treatment of heart problems can include medication, pacemaker, implantable cardioverter defibrillator (ICD).
- Respiratory aids: muscle training, assisted cough techniques, assisted ventilation etc.

Annual cardiac and respiratory assessments are recommended including ECG, Holter monitoring and/or echocardiography, respiratory function testing.

Triggering agents for malignant hyperthermia and volatile anesthetic medication should be avoided.

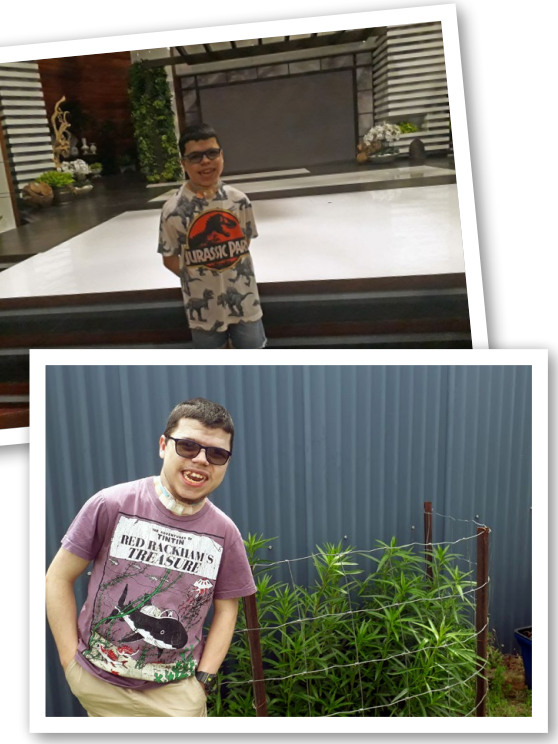
Relatives of people diagnosed with autosomal dominant EDMD and female carriers of X-linked EDMD should have cardiac evaluations as they may not have any other traits except heart issues.

The MDANZ Fieldworkers are available for support. They have in-depth knowledge and will have a good understanding of your needs and challenges. Ⓡ

A member's STORY

Life is there for living

Marcus Gower lives with Emery-Dreifuss muscular dystrophy and says when his diagnosis came, the "jigsaw" started falling into place - finally.



Top: Marcus Gower visits Ellen DeGeneres Studio in Hollywood. *Above:* Marcus shows off some of his swan plants.

Hi, my name is Marcus Gower and I am 20 years old.

I live in Pukekohe, Auckland with my Mum (Shona), Dad (Damon), brother (Vaughn) and sister (Chloe). We have two cats and two dogs.

My main hobbies are watching movies, playing PlayStation and spending time with my family.

I also have a bunch of swan plants so during the summer I spend hours watching the caterpillars grow. When they become a chrysalis and then hatch into beautiful Monarch Butterflies I feel like a proud parent when they fly off to start their new journey.

When I was five years old and started school, I was beginning to walk really high up on my toes and my muscles became really tight.

I ended up having an operation to lengthen my calves in an attempt to release the tightness. After the operation, serial casting, physio and stretching exercises they slowly became tight again.

One day when I had been in theatre for an airway scope the ENT surgeon had mentioned to Mum that my neck was getting hard to tilt back but couldn't, at the time, give a reason why.

A couple of years later I had a rib graft done to replace the floppy part of my airway but it failed and went floppy again. They tried again but it didn't work this time either. It was a mystery to as why this was happening.

It wasn't until after a routine MRI for something else and some blood tests that in February of 2011 when I was 10 years old, my pediatrician told Mum and I that I had a neuromuscular condition.

This was a bit of a shock, and we left our appointment feeling rather upset.

Later down the track things started to fall into place. Parts of the jigsaw were joining up… finally.

I was then confirmed to have Emery-Dreifuss muscular dystrophy (EDMD).

I live with a lot of different medical issues and although other people have the same issues, we are all on the same journey, but our own roads.

Having a tracheostomy tube (breathing tube) since I was a baby has had a huge impact on my life but with Emery-Dreifuss I do struggle a lot.

The main struggles are: general fatigue, getting up and down stairs or from a stool, dressing, showering, hand weakness and numbness, opening small bags such as chips or Lego.

In September 2017 I was shouted a trip to Hollywood where I got to see The Muppets live at the Hollywood Bowl. It was amazing!!!

One wish I have in life is to meet Ellen DeGeneres and while on a visit to Warner Brother's Studio our guide took us to her studio. I couldn't believe it, they had gone there just for me. Sadly, she wasn't there but it was still an experience I will never forget.

My advice to others living with this condition, or any other for that matter, is live your life because it is there for living.

Smile and stay positive, it can get you through a lot.

If you need to have time out, then take it.

If you need to rest, then rest.

If you need help, ask for it.

But most of all do all the things that make you happy and enjoy them.

Genetic testing fast evolving

Even if you have seen a geneticist in the past, there may be more information that is available to you today to help with a diagnosis, says clinical geneticist, Dr Gemma Poke.

If clinical geneticist Dr Gemma Poke could send just one message to MDANZ members it would be to say that genetics is changing increasingly rapidly so, even if you have seen a geneticist in the past, there may be more information that is available to you today to help with a diagnosis.

She explains that when she started her specialist training 11 years ago the tests available were much less sophisticated than they are today, noting that every year the testing gets better.

A clinical geneticist, she says, sees people who are without a diagnosis "and our job is to find a diagnosis for them".

As she describes it, she is a GP for rare diseases and says her team sees people from those not yet born to others well into old age.

They assess the clinical evidence, investigate the family history, closely examine the patient and work out the most appropriate genetic tests for that person. Once the testing is done, they interpret the results for the patient.

A typical appointment is an hour long and she says they need that time as family histories are complicated and they never know what tiny clue might lead to a breakthrough.

Gemma adds that as well as finding answers for people, another role clinical geneticists play is helping with education.

"Because we work with a lot of people with very rare diseases we can help educate a patient's own doctor about their disease," noting that they look to have an ongoing relationship with the patient and their doctor.

The service can also help those with rare conditions, who may be the only affected person in New Zealand, connect with others overseas.

Clinical geneticists are considered tertiary specialists, and a person will often have seen another specialist before they are referred, although if there is a strong family history a GP might refer someone directly.

As to the difference between a molecular geneticist and her role as a clinical geneticist, she says her role is a specialist doctor, while molecular geneticists are scientists. (See *In Touch* Winter 2021 edition for the story on molecular genetics.)

So does she find people are happy to get a diagnosis?

She says that getting a diagnosis can help relieve the uncertainty people have had for years, and may relieve any guilt parents may feel about what they did, or didn't, do.

She loves her job, and although it can be frustrating that treatment is lagging behind diagnosis, diagnosis has to come first to allow for targeted treatments.

Gemma works for the Capital & Coast DHB with a team of five clinical geneticists and six genetic counsellors.

The Wellington hub is one of three under the national Genetic Health Service NZ. It covers the lower half of the North Island and Nelson and provides outreach clinics in New Plymouth, Whanganui, Napier, Palmerston North, Porirua, Lower Hutt, and Nelson.

The Northern Hub, based in Auckland, covers the upper half of the North Island and provides outreach clinics in Whangarei, Hamilton, Tauranga, Rotorua, and Gisborne.

The South Island Hub, based in Christchurch, covers all of the South Island (except Nelson) and provides outreach clinics in Greymouth, Timaru, Dunedin, Queenstown, and Invercargill.

See www.geneticheathservice.org.nz.

Dr Gemma Poke is a clinical geneticist at the Genetic Health Service NZ, Central Hub in Wellington.

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 as 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.

National Support Team

Trevor Jenkin
National Chairperson

Natalie Foote
Executive Assistant

Brian Hadley
Accountant and Business Manager

TBC
Communications and Marketing Advisor

Kristin Cross
Fundraising and Partnerships Advisor

Shelley Butler
Accounts Assistant

Our branches

Northern Region

Fieldworker: *Mike Schneider* Office Manager: *Denise Ganley*
Ph: 09 415 5682 or 0800 636 787 Email: support@mdn.org.nz

Central Region

Community Co-ordinator – Wellington Region: *Pip McLean*

Fieldworker: *Talitha Vial*

Ph: 0800 886 626 Email: members.central@mda.nz

Canterbury Region

Fieldworkers: *Ross Paterson (left)* and *Jane Hazlett*

Southern Regions

Fieldworker: *Jackie Stewart*

Office Manager: *Vivienne Fitzgerald*
Canterbury: 03 377 8010 or 0800 463 222 Email: canterbury@mda.org.nz
Southern: 0800 800 337 Email: southern@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
Joy Jenkin
support@mdn.org.nz

Central Branch
Tristram Ingham
members.central@mda.nz

Canterbury Branch
TBC
chairperson@mda-canterbury.org.nz

Southern Branch
Trevor Jenkin
chairperson@mdn.org.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

COME ROLL WITH US AND SUPPORT KIWIS WITH MUSCULAR DYSTROPHY

WE'D LOVE YOU TO JOIN US!

Roll 1k For MDA

Annual appeal
20–26 September

To sign up or donate, visit
www.roll1kforMDA.org.nz

Can't participate? You can support someone who has already signed up, or click on the 'Donate' button to give directly to **Roll 1k for MDA**.