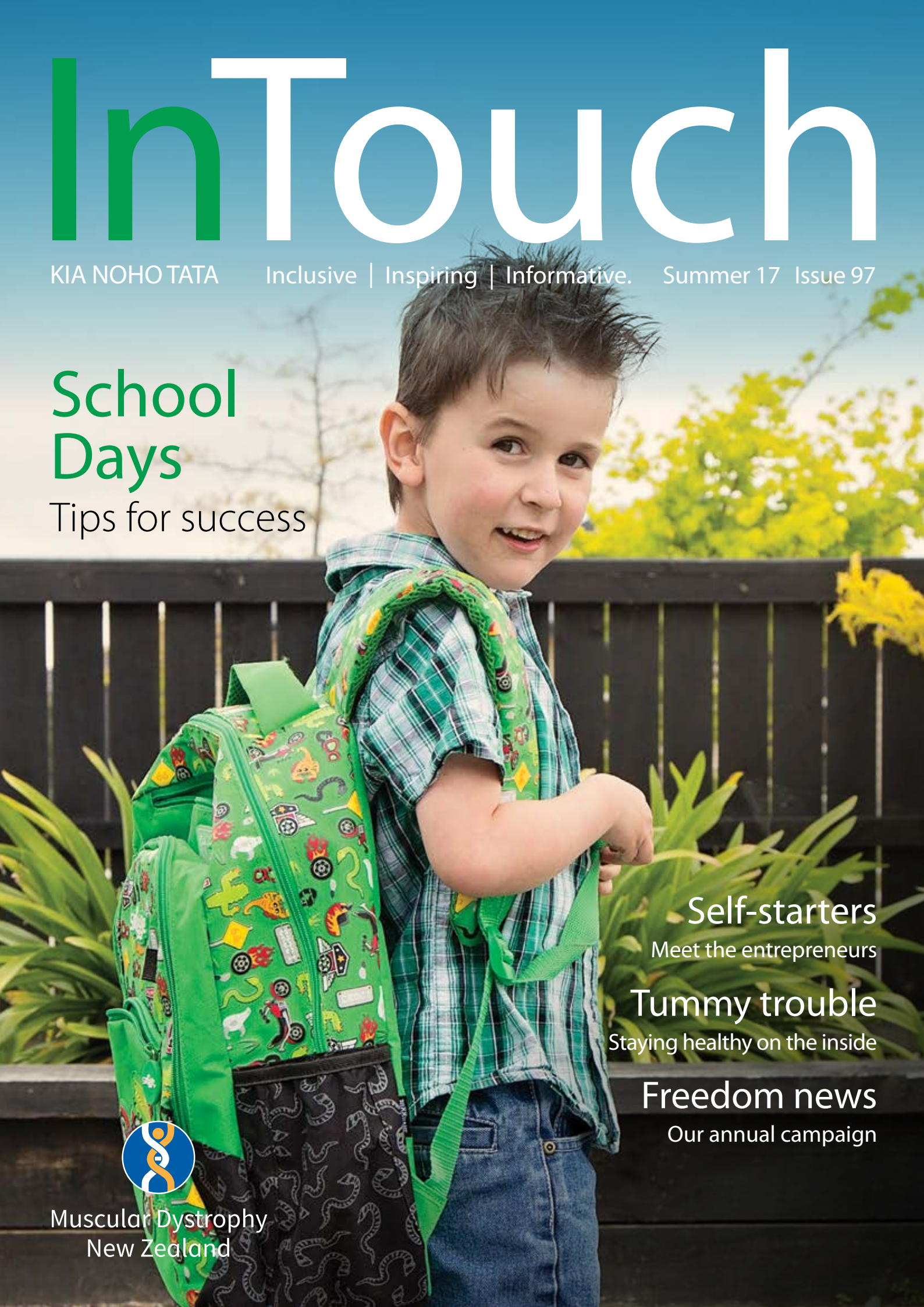
InTouch

KIA NOHO TATA Inclusive | Inspiring | Informative. Summer 17 Issue 97



School Days

Tips for success

Self-starters

Meet the entrepreneurs

Tummy trouble

Staying healthy on the inside

Freedom news

Our annual campaign

Muscular Dystrophy New Zealand

InTouch

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

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

Design: The Artset
the.artset@gmail.com

Cover photography: Gemma Foulds
Contributions: We welcome contributions, comments and letters to the editor. We thank all contributors to this edition. Contribution deadline for next issue: 22 January 2018.

Subscriptions: *In Touch* is available free to people with neuromuscular conditions, their families, health and education professionals and other interested people.

Advertising: *In Touch* welcomes advertising enquiries. For a rate card, please contact the editor.

Printer: Alliance Print
09 358 5151
allianceprint.co.nz

Charities Commission Registration:
CC31123
ISSN 1179-2116

Contents

FEATURES

Muscular Dystrophy New Zealand

MDANZ would like to thank the following supporters:

Lawrence Z. & Jacqueline A. Stern foundation
FOUNDATION NORTH
Lottery Grants Board
Pub Charity Limited
the Southern trust
Infinity Foundation Limited
Sir John Logan Campbell Residuary Estate
Nick Lingard Foundation
Disability Workforce Development
Four Winds Foundation Limited

REGULAR

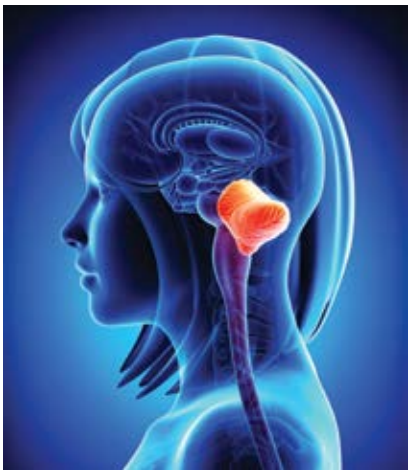
MDA NEWS

FREEDOM NEWS

BRANCH NEWS

RESEARCH

YOUR CONDITION IN REVIEW



GUEST SPEAKERS

ASK THE PANEL

We would also like to acknowledge our corporate sponsors:

horizonpacific · vega · CHAPMAN TRIPP · HYUNDAI

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

Korero from Ken

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

What a privilege it is to be part of MDANZ. I am continually reminded of some wise words that suggest we should not measure our lives by the breaths we take, but rather the moments that take our breath away.

Our Freedom campaign during September provided many 'wow' moments. Congratulations to the staff at MDANZ, the many hardworking volunteers, dedicated supporters, and – most of all – our members who have made our events so special. The dads who bungeed off the Auckland harbour bridge, the people who supported the movie night in Wellington, or the quiz night in Dunedin, all typify the efforts that led to successful outcomes.

I was fortunate to participate in two events. The first was a presentation by researchers focusing on facioscapulohumeral dystrophy (FSHD). Post diagnosis, we tend to lose sight of developments towards a cure, or means to stop progression of our conditions. It was heartening to hear of scientific developments by dedicated researchers. The learning point for me was making sure I was on the New Zealand Neuromuscular Disease Registry and to have a genetic diagnosis. Talk to your fieldworker or contact national office for more information on this.

The second event was the cocktail party fundraiser for our Duke of Edinburgh programme. What a great success! A huge thank you to those who gave their time, or items to auction. It was special to meet members and families, patrons and other special guests.

Life can be a challenge, but MDANZ wants to give you some 'wow' moments, and I thank you for giving me some in return.

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

Ken

Ken Green
MDANZ Chairperson

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.

Thank you. We greatly appreciate your support.

In touch with Ronelle

Tēnā koutou katoa, greetings to you all.

Whāia te iti kahurangi ki te tūohu koe me he maunga teitei. **Seek the treasure you value most dearly. If you bow your head, let it be to a lofty mountain.**

This whakataukī, or Māori proverb, is about aiming for what is most important to you, being persistent and recognising there may be obstacles to reaching your goals.

I thought of this proverb as I reflected on our wonderful Freedom campaign and on the vision of our organisation, *Freedom beyond limits*. My driver's license was recently up for renewal. Recognising changes to my strength and function, I went through a process of getting medical clearance and renewing my license for five years. Though it's hard to admit, I'm aware that I may need to stop driving before this term ends, for my own safety, and the safety of other road users.

Change such as this requires us to consider new possibilities and to plan. I'm starting to think of technology enablers that will help me stay independent for longer. Will a driverless car that I could programme and instruct using my voice be an affordable option? Will Uber for mobility vehicles be available by then? What if my work and home were located in close proximity and I could realistically travel between the two by power chair or using public transport? These are big questions for me to answer and I'm glad to have a strong network of informed friends to bounce ideas off, and to have enough foresight to plan ahead for a future where I am free to do the things that are important to me.

This kind of future thinking is what we have also been applying at MDANZ in the work we do and the systems and relationships we are building to strengthen the organisation. There are many external forces impacting the not for profit sector and we must continue to proactively plan for and respond to change. Our organisational values are the perfect compass to guide our direction. It will also take leadership, courage, persistence and foresight to take our organisation into the future.

We set some ambitious goals for 2017 and as the year ends, it is time to reflect on our successes, identify our priorities and organise our resources around these for the coming year. I thank all of the governance members, staff, supporters and volunteers who have contributed to another successful year for MDANZ and our four regional branches.

We hope you enjoy some contemplative time this summer, and that this issue of *In Touch* can add to that experience. In this issue, the team have compiled some stories about starting school, digestive health and fostering an entrepreneurial spirit.

I look forward to that entrepreneurial approach expanding the UberASSIST framework to better cater for those of us with power chairs. WiFi and carpool karaoke optional extras would be great!

Ngā mihi o te Kirihimete me te Tau Hou. Season's greetings for Christmas and the New Year.

Ronelle Baker
Chief Executive

A cup of tea and a catch up with … Kate Longmuir

Each issue we introduce a MDANZ team member:

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

In the past I volunteered at the National Office in Penrose and also spent six months as the Grants Fundraiser in 2015. Currently I am one of the Northern Branch Fieldworkers and I have been in this role since April 2017.

What qualifies as a great day at work for you?

One where I contribute to a good outcome, circumvent bureaucratic barriers and enjoy meeting new people. It has been a real privilege to meet MDANZ members over the past six months, to learn about the concerns and issues that people have and to be able to support people where necessary.

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

I would like to see members enjoying the activities that are meaningful to them and that provide a sense of purpose and fulfilment. I would like to see barriers to social participation removed and the amount of time that many members seem to spend negotiating bureaucracies reduced.

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

Delicious salads, spinach and feta filos, fruit kebabs, walnut crackers with pesto, cream cheese and sundried tomato dips. Yum! Good coffee essential.

What are you passionate about?

I'm passionate about social issues and social justice. I'm involved in a campaign to stop the logging and mining of a world heritage site in the Solomon Islands. Rennell has the largest fresh water lake in the Pacific and is the most beautiful, remote island. Recently an overseas mining company has started mining for bauxite. It is a social, economic and environmental disaster.

Christmas Hours

National Office and the branches will close for the holidays on **Friday December 23rd at 2pm** and reopen on **Monday January 8th.** Check mda.org.nz for emergency contacts during this time.

Preventing Pneumonia through Vaccination

More about our survey.

We ran a member survey in March 2017, asking about your experiences with pneumonia and to get an idea of how many members had accessed vaccination against pneumococcal disease.

Overall, members identified that more information should be provided about the pneumococcal vaccine for people with neuromuscular conditions as it can be a very serious illness and with a long recovery time.

There are two recommended pneumococcal vaccinations (Prevenar 13 approx $200 and Pneumovax 23 approx $70) which protect against different strains of bacteria that can cause pneumococcal disease and pneumonia. The criteria set by PHARMAC means that most of our adult members aged over 16years will not qualify for government funding of the vaccine.

Because cost is as barrier to having the vaccine, MDANZ has been offering to cover the cost of Pneumovax 23 for more than a year now. However we have only noticed an increased uptake of pneumococcal vaccines since running the survey in March. We have paid vaccination costs for 10 members in the past six months, ranging from 17 – 74 years and residing in various locations across the country. There are more males than females seeking the vaccine (8 vs 2).

MDANZ has also received several queries about funding for Prevenar 13 which is a much higher cost. In these instances, we encourage members to apply for additional funding through the Members' Discretionary Fund.

Queries continue coming in about pneumococcal vaccine funding, not just from members but from clinicians and specialists in hospital. We will continue our role as educators and provide information, to support people to improve their odds against getting pneumonia.

Our ultimate goal is to advocate for a system change and to have people with neuromuscular conditions covered under the criteria for government funding for both vaccines.

We submitted a formal application to PHARMAC in May 2017 and this application was on the agenda for the Immunisation Subcommittee of PTAC meeting, which took place on Wednesday, July 26th 2017. MDANZ is yet to receive minutes of the meeting and an outcome for the application.

Save the date

key dates for 2018

NEW YEAR 2018

AGM – Friday April 20th, National Office

Members Discretionary Fund Closing dates for funding rounds

- January 31st
- April 30th
- July 31st
- October 31st

September – Freedom Campaign

Our Dukies at work

Keeping up with their progress.

With the help of award leader Marty Price, our Dukies have begun working towards the Bronze level of the Duke of Edinburgh's International Award. They have set their goals – here's what they are working on.

Dylan Schneider, 14, from Auckland
Service: Dog petting therapy at retirement homes
Skills: Vex IQ robotics programming
Physical: Air rifle shooting

Ciaran Calder, 14, from Motueka
Service: Assisting with care of dogs
Skills: Rifle shooting
Physical: Table Tennis

Ella Mills, 15, Waiuku
Service: Volunteering at RDA (Riding for the Disabled)
Skill: Hairdressing
Physical: Badminton

Grace Chapman, 17, New Plymouth
Service: Making puzzles for hospice
Skill: Cake decorating
Physical: Gym workouts

Jack Lovett-Hurst, 20, Invercargill
Service: Radio show (assisting)
Skill: Cooking
Physical: Cycling

Good luck everyone!

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

MDA news

Australasian Neuromuscular Network

A small group of Kiwis made a big impact.

The Kiwi contingent; Ursula Gee, Ronelle Baker, Miriam Rodrigues and Miriam Hanna.

New Zealand was well-represented at this year's annual Australasian Neuromuscular Network's Scientific Meeting held at the Murdoch Children's Research Institute adjacent to Royal Children's Hospital, Melbourne in July.

Miriam Hanna, MDANZ's Information & Resource Co-ordinator presented her work on the pharmacological and non-pharmacological treatments used by people with myotonic dystrophy in New Zealand. **Ursula Gee**, Occupational Therapist at the Wilson Centre in Auckland shared with us families' assessments of the Paediatric Neuromuscular Management Clinic. **Ronelle Baker**, CE of MDANZ presented an overview of the outcomes found in the MD Prev study. **Miriam Rodrigues** provided a keynote presentation on neuromuscular research in New Zealand. She also presented work on the molecular diagnostic rates in New Zealanders with inherited muscle disease as well as a presentation on the attitudes towards reproductive carrier screening among this community.

Professor Nigel Laing spoke about reducing the burden of neurogenetic disease through population screening. He discussed the balancing act between prevention and therapy interventions, which are often expensive and unobtainable. A pilot providing reproductive screening for over 400 different conditions that can cause death by the age of four years is to get underway in Western Australia. As everyone is, on average, carrying genetic alterations for at least three to five recessive genetic disorders we all play "genetic roulette" when we have children. He stressed that reproductive screening can only ever lessen the burden but not eliminate it.

Vivienne Travlos, physiotherapist and PhD candidate from Western Australia, has been exploring well-being in youth with neuromuscular disorders who are wheelchairs users. She found that young people who are wheelchair users fare better and have increased wellbeing when they have friends and opportunities for socialising.

Dr Ian Woodcock is the Neuromuscular Fellow at Royal Children's Hospital Melbourne. He outlined the rationale for an upcoming clinical trial of creatine in children with facioscapulohumeral muscular dystrophy. Creatine is recommended for use in Duchenne muscular dystrophy where it appears to have a three-month peak response time increasing muscle force by 10%.

SMA community

We are looking for eight people with experience either as a parent or an adult with SMA to become part of an advisory group working with MDANZ on projects that will benefit our SMA community. Please contact Miriam Hanna if you are interested miriamh@mda.org.nz or phone 0800 800 337

NZ NMD Registry contributes to international study

Global analysis of DMD.

Sixty-four authors from 31 different countries, including New Zealand's Miriam Rodrigues and Richard Roxburgh, have published in the Journal of Neuromuscular Disease a detailed summary of clinical outcomes in the largest cohort of children and adults with Duchenne muscular dystrophy ever assembled. **Clinical Outcomes in Duchenne Muscular Dystrophy: a study of 5345 patients** is the partner publication to **The TREAT-NMD DMD Global Database: analysis of more than 7,000 Duchenne muscular dystrophy mutations** published in Human Mutation in 2015. The data was drawn from the TREAT NMD global network of Duchenne Patient Registries, including the NZ NMD Registry, and due to the statistical power afforded by the large numbers is able to answer questions about clinical outcomes that smaller groups of patients are unable to.

Thanks!

Your survey responses are being reviewed.

The trustees of Neuromuscular Research New Zealand Arthur Young, Richard Roxburgh, Leigh Hale, Graeme Hammond-Tooke, Alexa Kidd, Gemma Poke and Tristram Ingham would like to extend a sincere thank you to all those who contributed to the recent survey about Research Priorities for Neuromuscular Research New Zealand. Responses from 149 members are being reviewed by the trustees and will form the basis for their guidance to the National Council on what MDANZ's Research Priorities will be for the next three years.

Applications for funding

Neuromuscular Research New Zealand received four applications for funding projects for 2018. The results of the funding round will be presented in next quarter's *In Touch*.

Graeme Hammond-Tooke

Introducing…

Associate Professor Graeme Hammond-Tooke.

Graeme is a neurologist with a particular interest in neuromuscular disorders. He is based in the Dunedin School of Medicine and Dunedin Hospital and is a trustee of Neuromuscular Research New Zealand.

He became interested in neuromuscular disorders when training in neurology in Johannesburg, South Africa.

After immigrating to New Zealand, he became involved with the Muscular Dystrophy Association of New Zealand, and contributed for many years as Medical Advisor and then as a Patron. He has continued to be involved with the organisation and was a founding trustee of Neuromuscular Research New Zealand. He is also a current member of the NZ NMD Registry Oversight Committee, steering group member for the MD-Prev Study and sits on the Henry Kelsey Research Scholarship Committee.

Reach for the stars

Adopting our own piece of the night sky.

Did you know there are more than 100 billion stars in the Milky Way? And the Milky Way - which Earth is part of - is only one out of 100 billion more galaxies! Out of all those stars, there is one called *Freedom* which has been adopted by the Muscular Dystrophy Association of New Zealand.

We launched our Freedom Appeal in September at the Stardome Observatory in Auckland. The Freedom Appeal is all about raising awareness and funds so people living with neuromuscular conditions can live a life without limits, and what's more freeing than a star out there in the galaxy?

The evening began with drinks and snacks, before everyone headed into the theatre for the official naming of the star and to watch a show. Thanks to the face of our Month of Freedom, five-year-old Hazel, for doing such a good job of naming our star.

Olivia Shivas

Gretel Harvey-Smith gets to be an astronaut; the Peterson family enjoy the show; Oliva Shivas and Hazel Smith name our star.

A night to remember

Raising money for our young people.

The beautiful Tamaki Yacht Club on Auckland's waterfront was the venue for our first-ever fundraising cocktail party and charity auction. Our esteemed guests, Sir Richard Faull, longstanding MDANZ Patron Judy Bailey, and medical and community leader Dr Lance O'Sullivan were joined by a crowd of 90 supporters, MDA members, staff and friends.

The evening was a huge success and through the generosity of our guests and supporters, we exceeded our target and raised an impressive $15,000 from the event. All proceeds will directly benefit young people with neuromuscular conditions, enabling their participation in our nationwide, disability-friendly, Duke of Edinburgh programme.

Top: Sir Richard and Lady Diana Faull.
Above: Chris and Judy Bailey.

Information sharing

Getting together in Christchurch.

Around 25 Canterbury Branch members gathered at the Commodore Hotel, to hear from a range of speakers at an educational seminar co-hosted between the branch and national office. Internationally renowned neurologist Dr Gareth Parry talked about auto-immune conditions. Currently a research Professor at Nelson-Marlborough Institute of Technology, Dr Parry distilled complex information into readily understood terms for the audience.

ParaFed Canterbury Operations Manager Ken Sowden offered a glimpse into the world of elite Paralympic sport, while promoting the broader message of participation in sport and recreation for physically disabled people of all ages. And on behalf of the research team, Miriam Rodrigues presented findings from the Impact CMT study, which was funded by MDANZ's research arm, Neuromuscular Research New Zealand.

Speaker Dr Gareth Parry.

Our children's health

Information and advice for parents.

A dozen parents (and one set of grandparents) joined the MDANZ team at Waipuna Hotel in Auckland to hear information and advice that will assist them to better manage their child's health related needs. Families travelled from as far as Whangarei, Tauranga and Tokoroa to attend the educational seminar on the afternoon of 9th September.

Dietitian Sarika Coetzee shared insights into healthy eating and weight management for children with neuromuscular conditions. Respiratory specialist Dr Liz Edwards talked about warning signs to look for that signal breathing might be becoming an issue. Paediatric neurologist Dr Gina O'Grady discussed emerging treatments for conditions such as spinal muscular atrophy (SMA) and Duchenne muscular dystrophy (DMD) and Researcher and psychologist Kelly Jones presented information from the MD-Prev study that was gathered from child and participants. The formal programme of speakers was concluded with MDANZ Chief Executive Ronelle Baker and Tamariki member Dylan Schneider talking about the Duke of Edinburgh programme and why this new initiative will be valuable for our young members.

Miriam Hanna made a permanent pledge for freedom.

She did it!

A permanent reminder.

Redefining what it means to be a loyal employee, Information and Resource Coordinator Miriam Hanna made Freedom permanent by getting a tattoo representing the concept on her shoulder. She raised money for MDANZ through a Givealittle page.

"I was expecting quite a bit of pain and was really nervous, but I can honestly say, it was fine, "says Miriam. "The best thing has been being able to raise awareness by doing something I have always wanted. I really enjoy my job and am inspired by our members every day and their commitment to freedom beyond limits."

BRANCH news

Catching up with news from around the country

There were a few nerves on display for the harbour bridge jumpers, but Lance O'Sullivan still took a phone call!

Photos by Jacinda Boyd

Northern

Five very brave dads agreed to launch the Month of Freedom with a Bungy jump from the Auckland Harbour Bridge, coinciding with worldwide Duchenne Awareness Day on 7th September. Despite winds gusting up to 40 knots, they all jumped and helped to raise funds through the Muscular Dystrophy Northern (MDN) Give a little page. Many thanks to the dads, AJ Hackett Bungy for their sponsorship, and the supporters, members and friends who came along on the day.

The branch also hosted a Family Fun Day on the 9th September at Blockhouse Bay Primary School. There were plenty of fun activities for children and it was a good way to raise our profile and celebrate the start of the campaign. Thank you to Kuldeep and Yasha Singh for organising the event, and all the generous sponsors and supporters.

The next family camp is scheduled for 8th-10th December at Ngaruawahia Christian Camp. Places are filling fast so email denise@mdn.org.nz if you are interested in attending.

Tentative dates for Christmas parties are Sunday 19th November in Auckland and Sunday 26th November in Hamilton. Venues to be confirmed.

support@mdn.org.nz

It was a beautiful day for the Hastings Blossom Parade.

Wellington

Members of the Wellington regional branch made their debut appearance at the Hastings Blossom Parade, which was perfectly timed to be in the middle of the Month of Freedom. The younger members had a particularly good time and handed out stickers and balloons to children in the crowd. There are already plans to take part again next year, so let Napier based Fieldworker Penny Piper know if you are interested.

A special fundraising screening of *Victoria and Abdul* at Petone's Lighthouse Cinema was nearly a sell-out event and raised $1,000 for the branch. A huge thanks to the local businesses who supported the event.

Thanks also to the many volunteers who donated their time and took part in street collections around the region on the 28th and 29th of September.

Thanks also to the Ahern family who held an art auction to raise money and awareness for five-year-old Harper who has Duchenne muscular dystrophy (DMD), and making a donation to MDANZ as well.

office.mdawgtn@xtra.co.nz

Canterbury

The street appeal on the 29th and 30th of September was a great success for Canterbury Branch. Special thanks to the Dark Side Entertainers and other volunteers who helped collect money and to the Christchurch public who gave so generously.

The annual Canterbury Branch kids camp was run in the first week of the October school holidays and this year, had a super hero theme. Branch Committee members Bonny, Warren and Colleen, and a group of student volunteers ran a fantastic camp at Mt Hutt. The children built racing cars, made burners to cook pikelets, and went swimming and jet boating. Some amazing new friendships and memories were made.

On Sunday 10th December the branch is planning a Pop-up Wardrobe - a second hand clothing sale with items priced at $5 and under. They are keen to receive donations of washed clothing in good condition to help raise funds for the branch. Please Call Gemma on 03 377 8010 if you can help.

mdacanty@xtra.co.nz

Top: Thanks to the street collectors for their time.

Middle and above: Camp gave our kids a chance to try new things and make new friends.

Southern

The Southern Regions Branch launched the Freedom campaign by holding a hugely successful quiz night on the 18th September. Members and their families put teams together and the local community supported the event by providing raffle prizes.

Delicious home baking was on offer at the branch's High Tea, another successful event on the Freedom Month calendar. Speakers included Margaret Cardno from the HDC Advocacy Service, and MDANZ's Ronelle Baker and Miriam Rodrigues. The community provided items for a raffle, which included interesting artwork from local artist Tony Rowe. The children were entertained by balloonologist Pippity-Pop and local Senior Constable Aaron Smith, who bought along a police car so the children could honk the horns and flash the lights. This was definitely a hit with both children and adults!

A further Seasons for Growth group and seminar is planned for Dunedin and Southland in the coming months. Contact Fieldworker Jo Smith for further details, 027 509 8775.

joanne@mda.org.nz

Ronelle Baker, Jo Smith and Robbie Verhoef supported the High Tea.

Proud Mum Caroline with her son Sam, who has Duchenne muscular dystrophy. Photo credit: Gemma Foulds

Ready, set, go!

Tips for making school days, the best days

Starting school is a significant milestone that requires lots of preparation for families living with a neuromuscular condition.

When Caroline McFelin walks through the school gates with her six-year-old son Sam, it's a little bit like accompanying a celebrity.

"Sam loves school," she says. "He likes going to the playground, where all the kids line up to go on the seesaw with him, while his teacher aide holds and supports him. He has become quite the celebrity and all the kids come and greet us as we walk in the gate at school."

While Sam couldn't wait to join his older brother Noah at "big boy school", Caroline says she wanted to make sure he was really ready before his first day at their local primary school in Christchurch.

"The most difficult thing with Sam starting school was letting go," says Caroline. "We kept him at pre-school for an extra eight months until we thought he was ready. The school was very supportive and reassuring, and made it a very easy transition."

Getting ready for school started many months earlier. He had appointments with a speech therapist and physiotherapist, and there were visits to meet the Principal, Deputy Principal and teacher to discuss Sam's needs and his parent's aspirations for his education. They discussed access for the wheelchair Sam uses for long distances, and the help he would need using the bathroom and accessing the school environment.

The next step was a term of school visits three days a week, when Caroline stayed in the classroom to provide useful feedback for the teacher. During this time, Sam got

Her advice to other parents getting ready to transition their children into school is to make their voices heard.

to know his teacher aides who are now with him every day.

Sam is a big fan of structure and routine, so slotted into the existing classroom routines well. He leaves school at lunchtime every day so he doesn't get too tired.

Caroline says she's loved seeing Sam flourish socially at school.

"In the past he has preferred his own company, but since starting school he has made friends and loves joining in with activities and loves playing with other kids a lot more."

Her advice to other parents getting ready to transition their children into school is to make their voices heard.

"Ask lots of questions and express your concerns, "she says. "Be straight up. You know your child best."

MDANZ is here to help

Our fieldworkers work in the community providing personalised support and education for members with lived experience of a neuromuscular condition. They are available to visits schools and 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.

Northern Branch fieldworker Darian Smith says he is always guided by the wishes of the family when he speaks at their school. Some children like to be in the class while he speaks, others prefer to be absent.

"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," he says.

As well as fieldworker visits, MDANZ produces information booklets for schools. These are currently being revised and new versions will be available in early 2018. Here's an extract from one of our guides.

Top tips for teachers

From the resources for schools who have a student with Duchenne muscular dystrophy (DMD)

- **Students look up to you as a role model.**
  Modelling a positive attitude and support for your student with DMD helps other students learn how to be helpful, non judgemental, and react around disability in general. Their exposure to someone with physical access needs and the ways you normalise it within the school environment has a beneficial effect on them throughout their lives and impacts attitudes toward disability within society. You're doing great work!
- **Room placement**
  Place the student within the rest of the class, rather than at the back of the room so he feels included and not an outsider. Keep the aisles wide enough for him to move around, and interact or come and go independently.

Comfort Cough available for purchase or rental

Comfort Cough has been designed to assist people to clear bronchopulmonary secretions.

Comfort Cough makes positive and negative pressure and assists the natural cough.

This device is clinically proven to be effective and is very intuitive and easy to use.

A reliable cost effective solution and is ideal for community situations.

Please contact us for more information or if you would like a free trial.

solutions@breathingandmedical.co.nz

www.breathingandmedical.co.nz

Phone: 0800 335 333

breathing medical
RESPIRATORY TECHNOLOGIES

A team approach is helpful in assisting your student to thrive in school life.

- **Boost participation in activities outside and in class**
  Accommodate activities such as in P.E classes by letting him be a timekeeper or finding ways to include him in activities without being overly demanding or fatiguing. Encourage participation, if the student is not already doing so, in class in a way that does not make him feel small in front of the class. Reinforce him for doing good work, and making a contribution.

- **Have a multidisciplinary approach within the school**
  Keep open communication channels between the different supporting agents such as the occupational therapist, physical therapist, school counsellor, and/or teacher aide, so they're all aware of issues of concern that may arise and, more importantly, the solutions that have been developed. A team approach is helpful in assisting your student to thrive in school life.

- **Plan in advance**
  As your student grows older, he will develop new strengths and abilities, but at the same time he will loose other abilities as his muscles get weaker. Therefore it is important to continuously plan ahead of the changes that will occur for your student later in the year, and to build upon other strengths, talents and interests.

- **Keep parents updated**
  Parents want you to contact them about any concerns you may have about their child or if you need any assistance with a situation as it changes. It's better to raise issues earlier rather than later so they can be dealt with. In some situations, a broad whānau approach is appropriate, involving siblings in school life. Families are an integral part of the team, and open lines of communication are vital.

LEVO
THE EXPERTS IN STANDING

OFFERING THE LARGEST RANGE OF ANATOMICALLY CORRECT STANDING WHEELCHAIRS!

Each wheelchair has been designed with over 40 years of experience, research, development and user feedback. LEVO is proud to offer wheelchairs that combine seating and standing positions matched to your unique biomechanical dynamics with superior maneuverability, size, weight, aesthetics and adjustability. All models are reviewed and certified by internationally approved testing institutes.

Both full power and manual versions are available.

C3 WITH GYRO

SWISS DESIGN • INNOVATION • QUALITY

Euromedical Ltd | info@euromedical.co.nz | free phone 0800 **33 88 77**
www.**euromedical**.co.nz

Meet the entrepreneurs

Support for self-starters

MDANZ members are an innovative lot. This issue we're starting a series on entrepreneurs who are using their talents creatively and carving out a new niche for themselves.

Tegan Morris

Tegan Morris loves to travel, but isn't able to get on a plane as much as she would like. So instead of travelling the world, she has discovered a way to bring the world to her – and earn a little bit of money at the same time.

She lets a spare bedroom out to visitors on Airbnb and loves the chance to meet new people.

"The best part about doing this has been meeting such a variety of people and I have made a number of contacts around the world. It has also been a great way to add variety to my day-to-day life, which can at times be overly settled for my travel-craving personality," says Tegan.

"As far as a financial incentive for hosting travellers, I didn't see it as being a cash cow or that it would make big money, because I didn't think the space that I offer warranted it. But the income it could potentially offer was definitely a factor in deciding to give it a go."

While Tegan was keen on the idea right from the beginning, it took her family a little while to get used to the idea.

"When I first mentioned my interest in hosting visitors through Airbnb my family were a bit concerned around things like making sure everything was safe, and whether the costs of hosting people would be more than offset by the income. They suggested I just get a flatmate or boarder, but I had experienced that before and wanted the option of closing the room to paying visitors if I had family or friends visiting, or if I was going away or needed a break."

Tegan has developed good systems to make things work well for her, and has the support of Airbnb if there are any issues.

"I think the main thing is making sure I keep track of what is in my house so I know if things need repairing or replacing, or if something were to go missing. I have not had issues with this and the Airbnb company offer support if there are issues. The only other small challenge we have encountered a couple of times is the language barrier, but Google translate has helped us through these situations."

"As far as preparation goes, there isn't an awful lot to do apart from logical things like making sure that the room looks well-maintained, that you have a comfortable and decent quality bed, bedding and towels, as well as the support you will need to be able to look after the space and prepare in between visitors. It's good to be aware of your local area and what features could attract visitors and whether your expectations are reasonable for what you are hoping to get out of the experience, whether it be number of bookings, type of visitors, or income. Also, because it is a business, you will need to have an accountant or someone who can process your taxes so that you meet your IRD obligations."

Welcome messages on the blackboard at Tegan's home.

Tegan has plenty of practical advice for people considering doing the same thing as she is, but most of all, she says, be ready to enjoy the experience.

"The advice I would give is; know why you are doing it, recognise that if you have a good location and a fair price you are likely to get a good response and will need to decide how busy you want to keep your guest room. Don't forget to use the calendar feature to block dates when you aren't able to host, and enjoy the experience."

Tips for being an entrepreneur

Scratch an itch: The philosophy behind many a successful start-up is scratching your own itch. If there is a gap in the market for something you need, see if you can find a way to fill it.

Be a sponge: Your friends and family could well be full of helpful information on anything from helping you work on a small business plan to getting the word out about what you are doing. Find people who have done something similar to what you want to do and ask lots of questions.

Be brave! Starting something new is always a risk, but remember the age-old words – nothing ventured, nothing gained.

Get social: Facebook, Instagram and other social media outlets are a great way to let people know about what you are doing and to find customers if you have a product or service you would like to sell.

Keep it simple: It's great to have big dreams, but keep things small and uncomplicated to start with.

Be confident: Be brave! Starting something new is always a risk, but remember the age old words – nothing ventured, nothing gained.

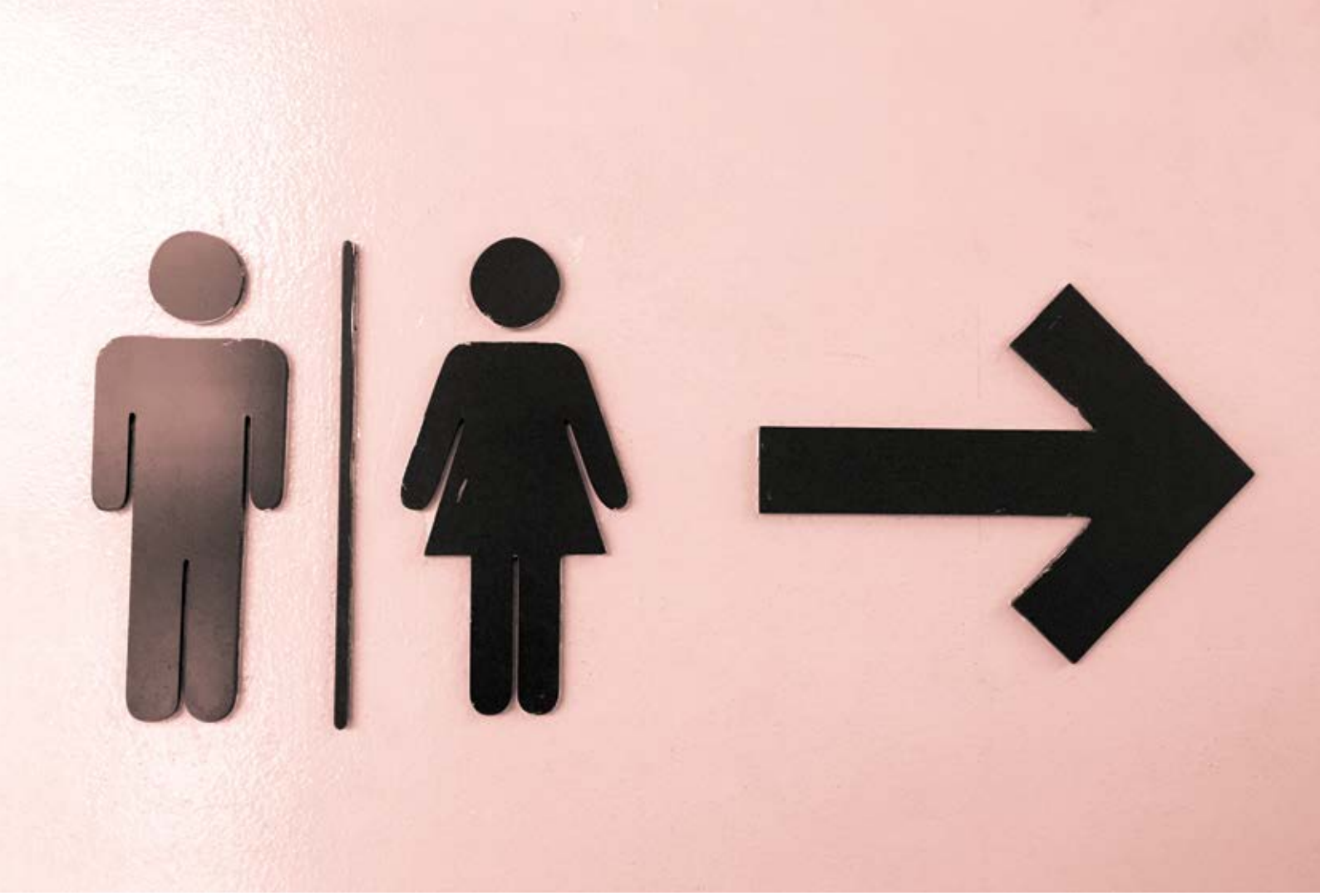
Where to get help

Check out these websites for practical help and support.

www.businessmentors.org.nz/Become-a-Client/Our-programmes.aspx

www.kiwiconnect.nz/entrepreneurs/

www.business.govt.nz/how-to-grow/getting-government-grants/what-can-i-get-help-with/



When you gotta go

Forget suffering in silence, let's get things out in the open. It's time for some toilet talk.

It's one of those taboo subjects that isn't easy to talk about, but here at *In Touch* magazine we're not afraid to tackle the tough stuff, so here's our guide to bowel health and neuromuscular conditions.

Because the symptoms of bowel trouble often come on gradually, it's easy to simply adapt to things being different to how they used to be, or dismiss them as not being important. But bowel health can seriously impact our overall health and wellbeing, so it's important to be vigilant, take any new symptoms seriously, and look after your digestive health.

How to maintain a healthy bowel

- **Keep your fluids up:** Aim to drink 6-8 glasses (1500-2000mls) of liquid per day – water is best. If you find it hard to drink plain water, try squeezing half a fresh lemon into a glass of warm water. It's tastier and provides extra nutrients, and is a particularly good way to start the day.

- **Fill up with fibre:** Include dietary fibre in your diet such as fruit (including dried fruit), vegetables, nuts, legumes (e.g. baked beans, kidney beans, soya beans), wholemeal breads and cereals.

- **Keep moving:** Regular exercise stimulates the bowels, so make sure you do some kind of movement each day. Where mobility is limited, you could try squeezing in your abdominal muscles several times a day while sitting and pelvic tilts while lying down.
- **Don't rush:** It's not the most exciting room in the house, but allow sufficient time on the toilet to get the job done. A regular routine is helpful, so try and go at the same time every day. The muscles in the stomach naturally contract after eating a meal, so you may want to take advantage of this process by having a routine after breakfast or dinner. It's important not to over-do things though, so rather than strain, leave after 20mins and try again another time,
- **Positioning:** Your daytime and eating posture is important, as being upright and supporting the stomach and spine will help digestion. Placing a stool in the toilet for lifting your leg height, can also help with bowel motions.

Treating the problems

Diarrhoea: This is most likely caused by reduced muscular activity (peristalsis) in the bowel, which leads to bacterial overgrowth. Medications that can help with this include Cholestyramine (Questran) and/or Norfloxacine (Noroxin). Domperidone or Metoclopramide can increase bowel motility.

Constipation: This is a common problem due to reduced mobility, and reduced fluid and food intake due to either chewing and swallowing difficulties, or lack of toilet access.

It's important to maintain your fluid intake, but it can also obtained from fruit and fruit juices, jellies, soups or yoghurt, which may be easier to swallow.

Introducing a food based product such as Kiwi Crush or Phloe to your diet, can help with every day regularity and these are available at supermarkets. Kiwi Crush can be diluted as a drink or made into frozen lollipops for children.

Sometimes it's hard to get enough fibre from diet alone, so Metamucil or Konsyl D can help, or laxatives may be required to achieve bowel motions.

There are numerous laxatives available which work in different ways. Some are softeners such as Lactulose syrup (Duphalac) and some are evacuants, such as Coloxyl Senna. Laxatives can be used in combination and adapted to individual requirements. The dose should be adapted to the lowest, most effective dose and the times of dosage experimented with to obtain the best result.

Your daytime and eating posture is important, as being upright and supporting the stomach and spine will help digestion.

Getting the posture right: Weakened abdominal muscles can affect the ability to sit correctly on the toilet and to bear down. This can be remedied by using equipment such as a toilet frame or footstool.

Severe constipation can occur with advanced stages of neuromuscular conditions, due to a reduced ability to bear down. Learning diaphragmatic breathing, or by blowing deeply or saying 'ommggge' when having a bowel motion, can push the diaphragm downwards, to help with evacuation.

Difficulty emptying (dyschezia), due to incomplete relaxation of the bowel muscles, causes a prolonged feeling of the urge to go, but an inability to pass a motion. Pelvic Floor Physiotherapists can help with this problem.

Incontinence: While neuromuscular conditions don't directly cause bowel incontinence, it can become an issue as the condition progresses. Reduced mobility and function can make getting to the toilet in time a challenge and then reduced fine motor dexterity can make getting clothes off in time an extra hassle. You may wish to experiment with different types of clothing that are easy and quick to remove, such as pants with elasticated waists and replacing buttons and zips with Velcro.

Tummy trouble: Gastric (stomach) problems caused by reduced stomach emptying include heartburn, reflux and regurgitation, which can lead to a serious symptom – aspiration of food or fluid into the lungs. Smaller meals often, with the main meal earlier in the day, can help with reflux at night. Taking time to chew your food and also having good posture and sitting at 90degrees or even standing while eating can help with this. Some people find dietary changes can help reduce gastric reflux, such as reducing coffee, chocolate, spicy food, acidic fruits, and alcohol. Aloe Vera juice and probiotics are natural products that may soothe the stomach and help with these symptoms.

Chronic constipation: This is a serious condition, which can lead on to impaction. A large mass of faeces can cause a dilated or mega colon and even bowel perforation in extreme cases. This is best treated with Lax sachets, also available in a children's dose, with maintenance treatment to prevent a recurrence.

An intermittent good clear out can also help prevent build up, using enemas or bowel washouts with equipment such as Peristeem Anal Irrigation system.

Children can develop chronic constipation or mega colon by holding on, or refusing to go to the toilet. Pain or difficulty having a bowel motion, can cause them to develop a fear of the toilet by association.

For severe cases, there is also a surgical option, a Caecostomy tube, (ACE procedure) for children and adults, where an enema is instilled into the bowel via a tube on the abdomen.

Aloe Vera juice and probiotics are natural products that may soothe the stomach...

If a person has a gastrostomy tube for PEG feeding, laxatives can also be given via this tube to help manage constipation.

Where to get help

It is important to ask for help if you have problems with your bowel function. Start with your GP, who may help directly, or refer you on to other health professionals.

Continence Nursing Service

Each DHB in New Zealand has a Continence Nursing Service, for pad supply or advice on bowel and bladder management.

Continence NZ can give help and advice via their confidential Freephone 0800 650659. They can also have contact details for the Continence Nurses throughout the country. Their website www.continence.org,nz is an excellent source of information and resources for adults and children.

Physiotherapists or Occupational therapists

For mobility needs, toileting equipment, and advice on exercise.

Dieticians For nutrition advice, modified diets, PEG feeding and dietary supplements.

Pharmacists For medication advice and information on probiotics and natural remedies.

Fostering connection with our members

Finding a nationwide community of support.

Connection and empowerment are two of our organisation's values, and that's what was on the agenda at these special events held around the country.

FA Family Day

History was made on the 15th of July, when 16 people with Friedreich's ataxia (FA) and their families gathered in the MDANZ offices. Never before in New Zealand has there been a gathering of this kind.

We were there to meet each other, learn more about our condition, and inaugurate the Friedreich's Ataxia Research Association New Zealand. This charity allows to fundraise in support of international research, which may include access to drug trials, and continue the efforts of FARA and MDANZ to create a specialist FA clinic in New Zealand.

Guest speakers at the FA day were Professor Martin Delatycki and Dr Louise Corben from the Melbourne FA research programme. A geneticist, Martin spoke in detail about current research, including the placebo-controlled trials of resveratrol underway to determine the therapeutic value of the drug. Resveratrol is derived from red wine, and Martin jokingly cautioned us against attempting to drink our way to a cure. A single therapeutic dose of resveratrol would mean having to drink hundreds of bottles in a single sitting!

Louise spoke more generally about the proactive approaches we can take to manage our symptoms day to day, and how we can stay healthy and active for as long as possible. Exercise is extremely important. Not only does it have benefits to improving strength and heart function, but is very important for mental health.

Studies have found that people with FA have higher rates of depression than the general population, which could be explained by the ongoing challenges and feelings of loss experienced simply by having FA. But,

The gathering at MDANZ National Office was one for the history books.

Louise mentioned there is another theory that postulates that the changes in the cerebellum of the brain from FA may be responsible. Ultimately, whether it is one cause or the other, the effect is the same. It is very important that people with FA have their mental health needs recognised and met.

From participants' feedback, everyone enjoyed the lively discussions during the afternoon brainstorming sessions discussing the challenges of living with FA and sharing ideas about what helps. It was the first chance we have had to be together in solidarity. May there be many more!

Joe Boon

FSHD Family Day

For the first time in its six year history, New Zealand was put on the map as the first stop on a traveling speaker series organised by Australian research based not for profit, FSHD Global.

Individuals and families travelled from Christchurch, Wellington, Tauranga and Hamilton to participate in

this unique opportunity held on 15th September at the Novotel Ellerslie, meeting other members with dacioscapulohumeral muscular dystrophy (FSHD) and hearing from scientific leaders in the field.

US based researcher Dr Scott Harper shared insights into gene silencing techniques as a potential treatment for FSHD. Starting the Harper Lab a decade ago, Scott is focused on developing RNAi-based treatments for dominantly inherited neuromuscular disorders like FSHD. Using a range of tools, including molecular techniques, viral vectors, and mouse models of disease, Scott offered hope that clinical trials may be only 3 years away.

This talk was complimented by Melbourne based researcher Dr Paul Gregorevic, who discussed his research programme at the Baker Heart and Diabetes Institute, Melbourne. Paul's focus on the mechanisms underlying the development and regulation of muscle has led to potential therapeutic interventions to combat muscle wasting and loss of muscle function associated with heritable conditions such as FSHD. This research will likely be beneficial to all people with muscle wasting conditions.

MDANZ chairperson Ken Green thanks the speakers

Dr Richard Roxburgh presented findings from the MD-Prev study and acknowledged FSHD Global for funding the analysis of information provided by those participants with FSHD. It was identified that 123 people have FSHD in New Zealand, geographically spread throughout the country, with most being adults and a slightly higher number of males than females. Many were in relationships, and while employment rates were slightly lower than the NZ average, many were in professional roles. While depression and anxiety rates were reported as lower than the NZ average by people with FSHD, their significant others (e.g. partners, parents) reported higher rates of depression and anxiety.

The SMA Family Day was such a success there will be another one soon.

SMA Family Day

Hosted at MDANZs National Office and held in collaboration with Julie Cini, Chief Executive of SMA Australia, an informal gathering to foster connection and offer information to individuals and families with spinal muscular atrophy (SMA) was held on 24th September.

With the emergence of Spinraza as a viable and safe treatment for SMA now being offered in parts of the world, the New Zealand community was keen to hear from Julie about the Australian experience of lobbying decision makers for registration and funding of the drug. While our countries clearly have different systems, learning from this experience and sharing information was very beneficial.

MDANZ Chief Executive Ronelle Baker talked with the group about the role of the organisation in supporting New Zealanders with SMA and the importance of a coordinated, collaborative effort towards lobbying initiatives. There are 75 known people with SMA in New Zealand, but without a prevalence study, the actual numbers in New Zealand and Australia are unknown.

MDANZ have met with Biogen, who is the pharmaceutical company involved in manufacturing and marketing Spinraza and there is a plan in place to register the medicine with Med Safe. It is exciting and helpful that MDANZ Programme and Service Advisor Miriam Rodrigues has been appointed chairperson of an international committee looking at the longer term monitoring and follow up of people who have been taking the drug, as the long term effects of Spinraza are currently unknown.

At the end of the afternoon, families expressed how much they enjoyed getting together and forming new relationships that will provide support for them beyond this experience. Plans are underway for another SMA Family day be held in Hamilton on 17th March 2018 in collaboration with SMA Australia and with a focus on well-being.

Another reason to drink your veges

The power of beetroot juice.

Researchers in the United States are interested to see whether beetroot juice extract could be helpful for people with Becker muscular dystrophy (BMD).

Beetroot juice is rich in molecules called nitrates, which are converted to nitric oxide in the body. The study will assess whether increased levels of nitric oxide produced in the body after consuming beetroot juice extract cause an increase in blood flow to muscle.

In the Phase 1 open label study, called "Skeletal Muscle Blood Flow in Becker muscular dystrophy," all participants will receive treatment with beetroot juice orally or via intravenous injection. Study investigators will assess whether the drug has positive effects on blood flow and handgrip strength. Researchers are currently look for people to take part in the trial, which is sponsored by Cedars-Sinai Medical Center.

Restless legs?

CMT, particularly CMT2 may be associated with Restless Leg Syndrome (RLS). People with CMT exhibit a threefold increase in RLS, which may result in increased fatigue, daytime sleepiness, and poorer sleep. Talk to your GP or neurologist about treatment options, and your physiotherapist about massage, exercise programmes or stretches and yoga. It is important to avoid potential triggers such as sitting in one position for too long, caffeine, tobacco and alcohol.

RARE DISEASE DAY 2018
28 FEBRUARY

The theme for Rare Disease Day 2018 is…

There is so much to be said in the fight to increase rare disease research that ***the theme for Rare Disease Day 2018 is research****, continuing on from Rare Disease Day 2017.*

Rare disease research contributes to the development of diagnostic tools, treatments and cures, as well as improved health and social care for people with rare conditions and their families.

More researchers are needed. They discover diseases and develop treatments and cures.

Researchers also need study participants and rely upon their participation to ensure research is meaningful, and translatable to clinical settings. Rare disease research is not done for the sake of creating knowledge; the knowledge generated is only useful if it is translated into real benefits for people living with a rare condition.

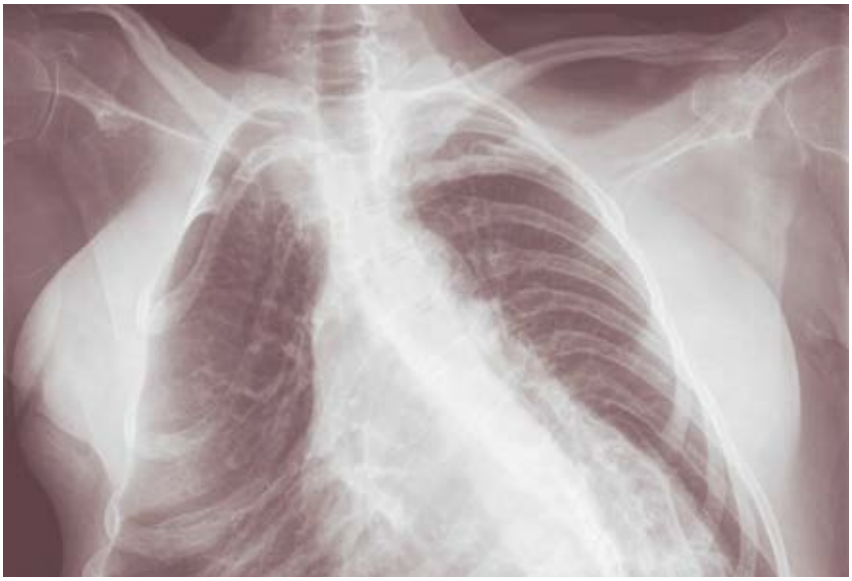
RAREDISEASEDAY.ORG

FDA Grants Orphan Designation to Mallinckrodt's MNK-1411 for DMD

Mallinckrodt Pharmaceuticals has developed a new drug, MNK-1411, for treatment of Duchenne muscular dystrophy (DMD). MNK-1411 is an injection composed of a synthetic hormone that activates melanocortin receptors and have been shown to have potential therapeutic effects on mouse models of DMD.

MNK-1411 has been recently granted orphan drug designation by the FDA, following Fast Track designation which was granted August last year. Orphan drug designation is a special status given to drugs that affect rare diseases, defined as having fewer than 200,000 patients in the United States. This will mean the company is able to exclusively market the drug in the U.S. for DMD following FDA approval for a period of seven years. The company can also use this program to apply for funding, tax credits for research costs, and eliminating the FDA application fee.

A Phase 1 clinical trial has been conducted for MNK-1411 and Mallinckrodt is analyzing the data obtained from that study to determine the optimal dose for a Phase 2 clinical trial set to begin in late 2017.



Spinraza continues to show positive outcomes

Study supports starting treatment earlier.

Spinraza (nusinersen) is an antisense oligonucleotide (ASO) that is designed to treat spinal muscular atrophy (SMA) caused by alterations in the Survival Motor Neuron (SMN1) gene that leads to survival motor neuron (SMN) protein deficiency. Spinraza results in increased production of SMN protein.

New data from the phase 3 clinical trial 'ENDEAR' has shown that earlier initiation of treatment with Spinraza improves motor function outcomes in infants with SMA, therefore the earlier it is introduced, the better the outcomes. The ENDEAR study is a randomized, double-blind, sham-procedure controlled 13-month study in patients with infantile-onset SMA.

Biogen's phase 2 clinical trial 'EMBRACE' also showed that infants and children treated with Spinraza were able to achieve greater motor milestones compared to those untreated. This study was a multicenter, randomised, double-blind, sham-procedure controlled 14 month study designed to assess the efficacy and safety of Spinraza in children with infantile- and later-onset SMA who were ineligible for other studies. The study also supports the dosing regimen of four loading doses in the first two months, followed by the administration every four months thereafter for infantile- and later-onset SMA.

Overall, results continue to reinforce the favorable efficacy and safety profile of Spinraza. The data was shared at the 22nd International Annual Congress of the World Muscle Society in Saint Malo, France in October 2017.

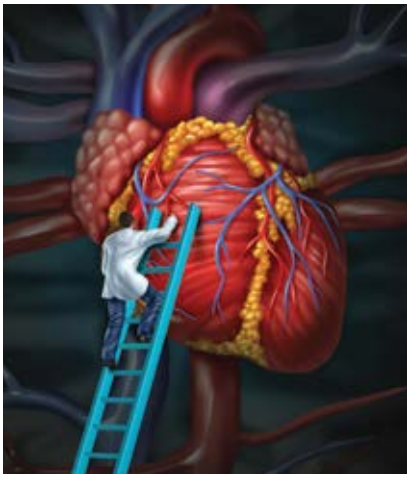
Capricor Therapeutics advances clinical development programme in DMD

Stem-cell based therapy.

Capricor Therapeutics which is developing biological therapies for Duchenne muscular dystrophy (DMD) and other rare diseases, recently announced that the national principal investigator for its HOPE-2 Trial will be Craig M. McDonald, M.D., a distinguished thought leader in the clinical management of neuromuscular diseases, including muscular dystrophies, and the development of novel outcome measures for DMD clinical trials.

The HOPE-2 Trial is being planned as a randomised, double-blind, placebo-controlled Phase II clinical trial that will evaluate repeat IV dosing of the study compound CAP-1002 which is a stem-cell based therapy, in boys and young men with DMD. Subject to regulatory approval, Capricor aims to start enrolling early next year.

Capricor Therapeutics also presented positive six-month results from the HOPE trial of CAP-1002, at the 22nd International Congress of the World Muscle Society, Oct. 3-7 in St. Malo, France. The study was designed to evaluate the safety and efficacy of a single dose of CAP-1002 delivered directly to the heart in boys and young men ages 12 to 25 whose DMD had involved their hearts. They were randomised into receiving either CAP-1002 treatment or usual care. The trial was open-label,



meaning the participants and the trial investigators knew who was getting which therapy.

At six months, thickening of the heart's left ventricle walls – which is a measure of the function of the left ventricle, crucial to pumping oxygenated blood to the body-increased significantly, by an average of 16.3 per cent to 31.2 per cent (depending on which part of the left ventricle was analysed) in the treatment group, while patients receiving usual care saw decreases in left ventricle wall thickness.

Performance of Upper Limb (PUL) tests also showed improvements in the middle-plus-distal regions (forearm and hands) in the treated group.

No difference in treatment-related adverse events was observed between the CAP-1002-treated group and controls who received standard-of-care therapy.

Capricor hopes to announce the results from the completed 12-month HOPE trial by the end of this year, although the study itself will continue through 2021. Ⓡ

Inherited ataxias

Understanding this group of genetic disorders.

Summary

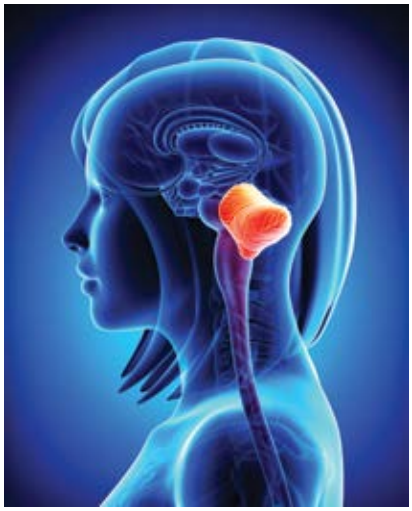
Inherited ataxias is an umbrella term for a group of rare genetic disorders characterised by slowly progressive incoordination of gait and often associated with poor coordination of hands, speech, and eye movements. Frequently, atrophy (wasting) of a part of the brain called the cerebellum occurs - the cerebellum is where movement, posture, and balance are coordinated.

MDANZ covers all types of inherited ataxias. These rare conditions include Friedreich's ataxia (FA), all types of spinocerebellar ataxia (SCA) and a condition known as CANVAS; cerebellar ataxia with neuropathy and bilateral vestibular areflexia syndrome.

Symptoms

There are many different types of hereditary ataxias and each may have unique signs and symptoms. However, in general, it is difficult to differentiate among the different types, and all are characterised by problems with movement that tend to get worse over time. Affected people may experience the following:

- Problems with coordination and balance (ataxia)
- Uncoordinated walk
- Poor hand-eye coordination



Wasting of the cerebellum frequently occurs.

- Speech difficulties (dysarthria)
- Involuntary eye movement
- Vision problems
- Difficulty processing, learning, and remembering information

Depending on the type of condition, signs and symptoms can develop anytime from childhood to late adulthood. Over time, the affected nerve cells begin to function poorly and ultimately degenerate. As the disease progresses, muscles become less and less responsive to commands from the brain, causing coordination problems to become more pronounced. Those affected by poor coordination will notice poor balance when walking, inability to run, clumsiness of the hands, a change in speech, or unusual eye movements.

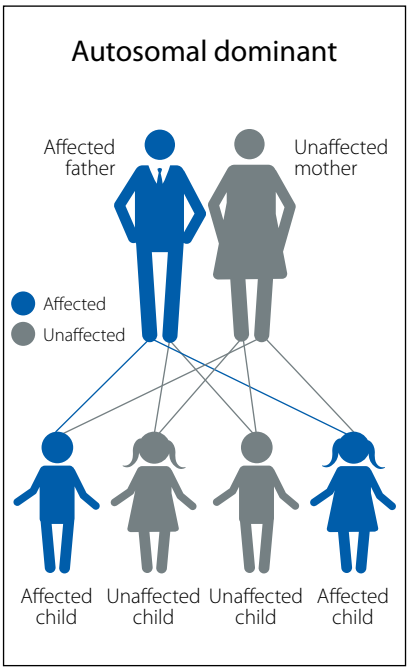
Individuals with inherited ataxias may develop a variety of other symptoms as their condition progresses, such as: numbness, tingling, or pain in the arms and legs (sensory neuropathy); uncontrolled muscle tensing (dystonia); muscle wasting (atrophy), muscle twitches (fasciculations), rigidity, tremors, seizures, tinnitus, vertigo, and involuntary jerking movements (chorea). The condition may be complicated by vision disorders and eye movement paralysis, or have association with heart disease, breathing problems, bone abnormalities and diabetes depending on the type.

FA is associated with cardiac problems, depression and type 2 diabetes. Cardiac complications include cardiomyopathy, myocardial fibrosis, heart failure, tachycardia or heart block. Many of these symptoms can be treated with medication.

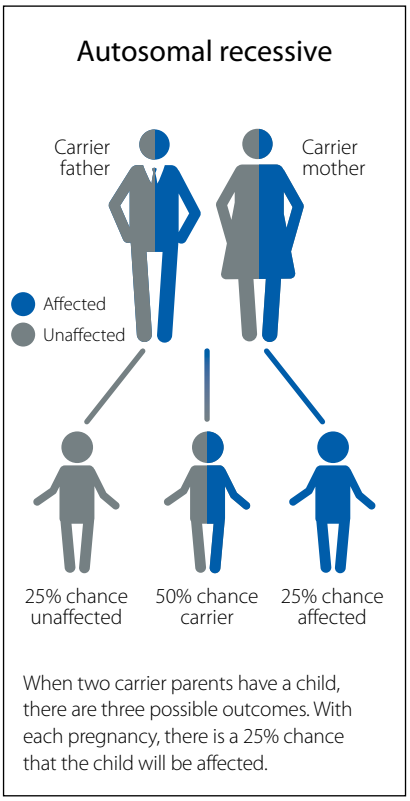
CANVAS is associated with a dry cough.

Cause

Mutations in many different genes are known to cause the different types of spinocerebellar ataxia (SCA). For some types, the gene known to cause it has been identified, while in others, the genetic cause is still unknown (about 40 percent to 25 percent of the cases).



Mode of Inheritance for the majority of SCA's (autosomal dominant inheritance). Also Friedreich Ataxia and some SCA'S (autosomal recessive inheritance).



When two carrier parents have a child, there are three possible outcomes. With each pregnancy, there is a 25% chance that the child will be affected.

In 1863, Nikolaus Friedreich (1825-1882), a German pathologist from Heidelberg, described a new spinal disease for the first time, which came to carry his name 'Friedreich's Ataxia'. It took a 120 years to discover the genetic defect underlying Friedreich Ataxia (FRDA) in 1996 named 'frataxin'.

The cause of CANVAS is currently unknown – this condition was only described in 2011 and work has been underway ever since trying to find the genetic cause of it.

Inheritance

Inheritance can be either autosomal dominant as in many of the SCA's, autosomal recessive as in FA or X-linked recessive. The inheritance pattern of CANVAS is yet to be fully understood but is considered likely to be autosomal recessive.

For some types of SCA and for CANVAS, the genetic cause is still unknown.

Diagnosis

A diagnosis of hereditary ataxia is often suspected when certain signs and symptoms, such as a poorly coordinated gait (walk) and uncoordinated hand/finger movements, are present.

Genetic testing is the best way to confirm FA and SCA and identify the specific type, however, this is only an option if the disease-causing gene for that particular condition or sub-type of condition has been identified. Genetic testing can confirm a clinical diagnosis or it can predict that an individual is likely to go on and develop the condition. Testing is available for many different genes known to cause spinocerebellar ataxia (SCA) and is also available to test for FA. Carrier testing for at-risk relatives and prenatal testing are possible if the disease-causing mutations in the family are known.

For some types of SCA and also for CANVAS, the genetic cause is still unknown. Genetic testing is not available for families with these types of conditions until the genetic cause is identified. In these cases, imaging studies such as computed tomography (CT scan) and/or magnetic resonance imaging (MRI scan) may be helpful in establishing a diagnosis. A CT scan is an imaging method that uses x-rays to create pictures of cross-sections of the body, while an MRI scan uses powerful magnets and radio waves to create pictures of the brain and surrounding nerve tissues.

Treatment

There is no known cure for any types of hereditary ataxia. The best treatment options for SCA and FA vary by type and will depend on the signs and symptoms present in each person. The most common symptom of hereditary ataxias is ataxia (a condition in which coordination and balance are affected). Physical therapy can therefore help strengthen muscles, while mobility aids (e.g., walking stick, walker, or wheelchair) can

CANVAS is associated with a dry cough.

Many will eventually need assistance to perform daily tasks.

assist in mobility and other activities of daily life. Many people with SCA have other symptoms in addition to the ataxia such as tremors, stiffness, muscle spasms, and sleep disorders; medications or other therapies may be suggested for some of these symptoms. One report described some improvement in the symptoms with zolpidem 10 mg in four out of five family members with SCA type 2, and a trial of 20 patients with SCA3 found that varenicline led to improvement in some, but not all of the symptoms.

Prognosis

The long-term outlook (prognosis) for people with CANVAS, FA or spinocerebellar ataxia (SCA) varies.

Most available information on the prognosis of SCA is based on the four most common types: SCA1, SCA2, SCA3 and SCA6. People affected by one of these types of SCA usually require a wheelchair around 10-15 years after the onset of symptoms. Many will eventually need assistance to perform daily tasks. Similarly the prognosis of FA is reasonably well described and is constantly being updated as more research is carried out.

Research

Research helps us better understand diseases and can lead to advances in diagnosis and treatment.

Clinical Research Resources

The US government website 'Clinicaltrials.gov' lists all clinical trials and some other research studies. Enter the condition that you're interested in into the search bar and then click on the generated list to read descriptions of these studies.

Please note: *We strongly recommend that you talk with a trusted healthcare provider before choosing to participate in any clinical study.*

Patient Registry

The NZ NMD Registry covers all neuromuscular conditions including all forms of SCA, CANVAS and FA. Talk to your fieldworker or contact info@mda.org.nz to find out more.

My condition doesn't stop me

Sue Hay shares her journey with CANVAS, how she learnt how to look out for herself, and accepted it's ok to rely on a friend now and again.

I grew up in Tauranga, one of five siblings. I had a normal, happy childhood, with lots of great memories. I don't recall noticing any of my family members with symptoms of neuromuscular conditions, not my parents or grandparents, or any aunties or uncles.

I married and had two children, Sarah who's now 32 and Bradley, now 29.

I have recently retired at 65, after 20 years working for Life Pharmacy and Farmers as an Estée Lauder Consultant, a job I really enjoyed, and was lucky enough to have supportive colleagues and managers over the years.

About 20 years ago, I joined my husband playing golf. He noticed that one day I was playing just fine, the next I wasn't, and there was something wrong with my balance. I went to my GP, who was a dear friend, and said I couldn't stand on one leg. Her response was, "Of course you can Sue". I convinced her I was serious, so she referred me to a neurologist.

He diagnosed sensory peripheral neuropathy, and all was fine for about eight years until I noticed a deterioration. Every time I turned my head, my eyes would lag behind and weren't quite following my movement. By then my GP had retired, so I went to a new one, who referred me to another neurologist in Auckland to investigate the deterioration.

After coordination tests, assessment of eye movements, and nerve conduction studies and scans, I was diagnosed with Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome (CANVAS). The neurologist referred me to Richard Roxburgh, who I'm currently seeing. Richard was running a research study and I had genetic testing done, unfortunately researchers are still trying to find the causative gene for such a rare condition.

My sister Judith, who is four years younger than me, then got diagnosed with the same condition. No one else in the family seems to have it, nor do our children, who could potentially be carriers.

The next stage was I started needing to see my feet to walk properly, as there was a lack of coordination in signals between my feet and my brain. It was the same with my arms and I couldn't always pick things up. After a few bumps and falls, I started to analyse the situation and investigate what I could do to keep myself safe. It's particularly hard to walk in the dark or rain. That's when I have to rely on a friend.

I try and keep a positive outlook on life. I don't let my condition stop me. I love gardening, cooking and reading – which I can still do. I'm fortunate to have a supportive husband and a son living just down the road. I drive small distances locally, and volunteer most days at the local Salvation Army store.

I've got an exercise bicycle at home which I use to keep my legs moving, and may join a gym to lift weights. I've had to give up swimming as I have no spatial awareness. I can't walk along the beach either as I need a firm surface.

Sue has retired, but keeps very busy.

I still travel, but can't do so on my own as I have to have someone with me. We recently did a lovely trip to the South Island, and went on the Trans Alpine Railway.

At home I have put rails in the bathroom and have an emergency button in case I fall and no one is around. Recognising what I needed to do to help prevent falls, means I haven't had one for a number of years

The best advice I can give people with ataxias is don't be afraid to ask for help and accept it when it's offered. Also, don't be afraid to tell people about your condition. It gives them a better understanding of how to help you better.

GUEST speakers

The cocktail party fundraiser was a great event.

Getting connected

OLIVIA SHIVAS

For me, the highlight of being Youth Rep for MDANZ is connecting with young people and meeting their families.

As part of our Freedom Appeal in September, I attended the cocktail party fundraiser which raised money for young people living with neuromuscular conditions to participate in the Duke of Edinburgh's Hillary Award. The Duke of Edinburgh programme is an individually-tailored programme to build skills, identity and self-esteem; there are four parts consisting of voluntary service, learning a skill, a physical activity component and an adventurous journey.

This is an exciting initiative and I know personally that something like this can have a huge impact. When I was 18, I completed the Queen's Award. It was a two-year project consisting of voluntary work, giving a speech at Parliament in a committee room in front of a MP, an exam about New Zealand governance, and organising events in the community.

I'm looking forward to following the progress of our Duke of Edinburgh participants and seeing them succeed!

To be honest, there were times when I considered giving up on completing the Queen's Award. I had lots of school work to keep on top of and extracurricular activities such as violin practice and singing. I'm a perfectionist, so I put a lot of pressure on myself to get top marks. However, I had encouraging parents and mentors who supported me through it – and it was so worth it in the end! My family and I went to the ceremony at Government House in Wellington and I received my certificate from Lady Janine Mateparae, the wife of the Governor General at the time Sir Jerry Mateparae.

Doing this programme gave me new skills and a boost of confidence as to what I could achieve. I'm looking forward to following the progress of our Duke of Edinburgh participants and seeing them succeed!

As always, I'd love to hear from other young MDANZ members; you can email me at olivia.shivas@gmail.com to get in touch. Or if social media is more your thing, we've just launched a private Facebook group for all members between 16 – 25 years of age. As we have young members living all across New Zealand, this is one place where we can come together and discuss matters that are important to us – such as starting university, getting a modified car to drive, or planning an OE. Request to join by searching 'MDA Youth New Zealand' on Facebook or scan the QR code below. Look forward to connecting with you!

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.

Bacteria: the good vs the bad

MIRIAM HANNA

Q: Prebiotics, probiotics and symbiotics – I get so confused! What are they and what's the difference?

It's true, we are made up of more bacteria cells than human cells in our body. Research has shown that the human gut alone contains billions of bacteria cells of varying strains, and that they're there for a reason.

Whilst people think of bacteria as harmful, infection-causing microorganisms that should not be present, probiotics are friendly beneficial bacteria, that may help to remove infectious bacteria and also prevent yeasts and fungi from growing excessively. Hence they are called Pro (for) biotics (life) instead of anti (against) biotics (life). Having a healthy gut does not mean no harmful bacteria, it means maintaining that balance, or ecosystem so that harmful microorganisms do not take over.

The effects of probiotics are strain-specific, which means that certain strains may be beneficial for a particular condition but not for another, so it is important to discuss with an expert.

Our gut flora has a huge impact on our lives and health, it plays an important role in our immunity, mood, weight and even mental health.

However, the composition of the gut flora varies hugely from person to person. There are gender related variabilities, and even race related variabilities if you consider the different diets people eat around the world. It's also important to note that babies are born with very little bacteria in their guts, and this starts to grow, multiply and diversify over time. So what is introduced to a child in the first 1000 days of life can impact their health outcomes later on in life.

If you are considering supplementation, you may get further advice from a GP, dietitian or a nutritionist. Health stores and Pharmacies are also helpful.

Probiotics are live bacteria strains, which is why most of the brands you buy need to be kept in the fridge – heat tends to kill bacteria and they become ineffective. The effects of probiotics are strain-specific, which means that certain strains may be beneficial for a particular condition but not for another, so it is important to discuss with an expert. Ferments that are commercially available, such as kefir, miso, sauerkraut, and high quality plain live acidophilus yoghurt are good sources of probiotic strains.

Prebiotics on the other hand, are plant fibres, found in common foods we eat such as onion, garlic, leeks, asparagus, raw unpeeled fruits and vegetables, plain nuts, seeds, and dried fruits, pure unadulterated dark berry juices, legumes, chickpeas, lentils, peas, oats and cashews. They nourish the good bacteria in our guts. Unlike probiotics, fibres are stable in formulations and not affected by heat or bacteria.

There might be times you consider taking probiotics – in winter when your intake of raw foods is low or you get sick often, if you are under stress or pressure, you have undesirable bowel symptoms, if you have allergies or food intolerances.

While probiotics introduce good bacteria into the gut, prebiotics act as a fertiliser for the good bacteria that's already there. When the two are manufactured in combination, they are now being referred to as symbiotics.

Miriam Hanna is Information and Resource Coordinator at MDANZ and is a practising and registered community pharmacist.

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 core team

Ronelle Baker
Chief Executive

Miriam Rodrigues
Programme and Service Advisor

Brian Hadley
Accountant and Business Manager

Miriam Hanna
Information and Resource Coordinator

See more about our team at www.mda.org.nz

Northern Branch


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

Wellington Branch


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

Canterbury Branch


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

Southern Branch

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

Council Representatives

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

Northern Branch

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

Wellington Branch

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

Southern Branch

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

Canterbury Branch

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

Conditions covered by MDANZ

Muscular Dystrophies:

Becker Muscular Dystrophy

Congenital Muscular Dystrophies and Congenital Myopathies

Distal Muscular Dystrophy

Duchenne Muscular Dystrophy

Emery-Dreifuss Muscular Dystrophy

Facioscapulohumeral Muscular Dystrophy

Limb-Girdle Muscular Dystrophy

Manifesting carrier of Muscular Dystrophy

Myotonic Dystrophy

Oculopharyngeal Muscular Dystrophy

Diseases of the Motor Neurons:

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

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

Type 2 Intermediate Spinal Muscular Atrophy

Type 3 Juvenile Spinal Muscular Atrophy (Kugelberg Welander Disease)

Type 4 Adult Spinal Muscular Atrophy

Hereditary Spastic Paraplegias (HSP)

- all types:

Also called Familial Spastic Paraparesis

Leucodystrophies

- all types.

Metabolic Diseases of muscle *- all types including:*

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

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

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

Phosphofructokinase Deficiency (also known as Tarui's Disease)

Phosphorylase Deficiency (also known as McArdle's Disease)

Diseases of the Peripheral Nerve:

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

Dejerine-Sottas Disease (CMT Type 3)

Hereditary Sensory Neuropathy

Inflammatory Myopathies:

Dermatomyositis

Inclusion Body Myositis

Polymyositis

Diseases of the Neuromuscular Junction:

Congenital Myasthenic Syndrome

Lambert-Eaton Syndrome

Myasthenia Gravis

Myopathies *- all types:*

Andersen-Tawil syndrome

Central Core Disease

GNE Myopathy

Hyperthyroid Myopathy

Hypothyroid Myopathy

Myofibrillar myopathy

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

Myotubular Myopathy

Nemaline Myopathy

Paramyotonia Congenita

Periodic Paralysis

Inherited Ataxias:

CANVAS

Friedreich Ataxia (FA)

Spinocerebellar Ataxia (SCA)

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

Central Cavernous Hemangioma

Neurofibromatosis Type 1

Neurofibromatosis Type 2

Schwannamatosis

Tuberous Sclerosis

Von Hippel Lindau Syndrome

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

Get out there!

molift Smart 150

Take with you anywhere

- World's lightest folding portable lifter at only 24kg
- 50% lighter than others
- Folds in seconds, stands up for compact storage
- Available with a wheeled transport bag

Seatara WheelAble

Be Spontaneous

- Lightweight at only 12kg
- Unfolds in 5 seconds
- Ready for use in the shower and over the toilet
- Available with a wheeled carrybag, soft-seat & commode bucket

VICAIR ALLROUNDER O2

Designed for adventures and activities outside of your wheelchair.

- All new design
- Available in 3 sizes
- Cooler - 100% breathable
- Lightweight - only 0.8kg
- Machine washable

Morton & Perry
Healthcare Equipment Solutions

www.mortonperry.co.nz 0800 238-523 advice@mortonperry.co.nz