InTouch

KIA NOHO TATA Inclusive | Inspiring | Informative Summer 20 Issue 108

A time to renew and rejuvenate

Smiling into the storm:
Coping mechanisms we can all use

★

Lighting up the world:
Through the magic of Aaron's computer

★

NZ road-trip:
4,000 kilometres of inspiration

Muscular Dystrophy New Zealand

InTouch

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Contributions: We welcome contributions, comments and letters to the editor. We thank all contributors to this edition.

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 MDANZ office.

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

Charities Commission Registration:
CC31123
ISSN 1179-2116

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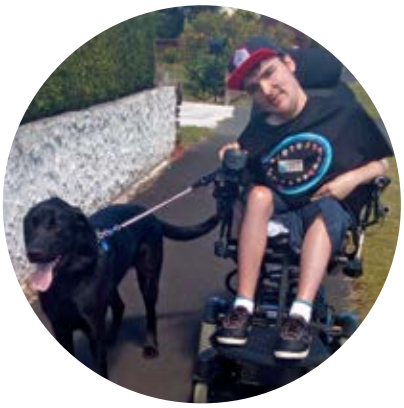

Muscular Dystrophy New Zealand

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Also thanks to Allied Medical, 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.

Kōrero with Tristram

A year to notice, savour and enjoy the small everyday things that happen to us while we are waiting for our lives to happen.

This year has been exhausting and a real roller-coaster for so many of us. Certainly Covid-19 and the pandemic has dominated the year and significantly impacted most aspects of our lives.

As I write this article in late spring, having still largely been in confinement – and somewhat hesitant to get back out in the community – I have been grateful for the spring blossoms that have been on the cherry trees outside my window, a friendly local tui, the return of warmer weather, and visits from my new grandchildren. I mention these because this year has taught me to try and notice, savour, and enjoy those little everyday things that happen to us while we are waiting for our lives to happen.

I've concluded that this philosophy is something of a metaphor for MDANZ. We all hold hope for the eventual cures of our neuromuscular conditions, and there are certainly a number of exciting developments in the pipeline, but to me the real role and blessing of MDANZ is the fellowship of our community, and the mutual support that provides, as we continually learn (and re-learn) to cope with our conditions and the impacts they have on our lives. As an Association we need to embrace, support, and celebrate those little everyday things that make a difference in our members' lives.

For most of our members, this year has been tough, and we recognise that many have come through it feeling bruised and battered. These feelings are common, it's healthy to reach out for help, and by working together and supporting one another we can build our resilience. The more examples and stories I hear of our members, how you cope, survive, and thrive, the more I realise that you are all remarkable people.

Like the season, this edition of *In Touch*, is about new renewal, the promise of fairer weather ahead, and growth. You will see that as an Association we are going through a period of renewal and development. We have seen a rapid increase in demand for our services, and a need to expand our roles in providing support for mental health, digital connectivity, and career transition advice.

I am proud of our hard-working team (staff, volunteers, and committees) up and down the country who have coped with disruption, changes to our business processes, working remotely, and feeling isolated, and all the while working hard to provide support for our members.

It's in the midst of hardship that we often see the best of humanity. MDANZ is like a family and it's the acts of kindness, connection and support that members have been showing each other that make this Association so unique and special.

On behalf of all of us at MDANZ, I want to wish you a holiday where you get to savour and enjoy the best that life has to offer - a relaxing fun time, full of joy (and Christmas pudding), surrounded by those you love.

Dr Tristram Ingham
National Executive Chairperson

MDANZ staff on the move

MDANZ has said "see you later" and "good luck" to some valued team members in the past few months, while we are also delighted to welcome new staff to the team.

Our administration and fundraising assistant **Amelia Noyes** left in August after joining MDANZ in October 2018. During her time with MDANZ, Amelia was responsible for running the website and assisting the team with delivering information to members. Amelia's strong passion for youth development saw her join the Duke of Edinburgh Awards Team as an Award Leader. We thank Amelia for all her contributions and wish her well for the future.

Paul Graham was a Fieldworker for the Canterbury Branch from July 2011 until he retired in September. Paul's empathy and assistance helped make positive changes to hundreds of members' lives over the years. Paul always put 110 percent into his role and is missed by members and staff. Paul always had his phone on and was always available for members. We wish Paul all the best for his well-deserved retirement.

Natalie Foote began working with MDANZ in November 2018 in a self-employed capacity. In July 2019 she joined the staff as the Executive Assistant and Marketing Executive in the National Support Office. Natalie rapidly became an invaluable asset and turned her hand to multiple things. She was a central point of communication and was the glue that held many things together. Natalie left in October ahead of the birth of her second child. We wish Natalie and her family well.

Fieldworker **Rachel Woodworth** was part of MDANZ's Northern Region for two and a half years before leaving the organisation in November. Rachel always had members' best interests at heart and put in a lot of work for them. She was known to be kind,

caring, considerate and supportive. Her dedication and commitment will be missed. The Northern Branch Executive Committee thanks Rachel for all she has done and wishes her the very best.

Meanwhile, we have welcomed **Raygaana (Ray) Naidoo** who began her role as the Greater Wellington-based Fieldworker for the Central Region in August. Ray supports members from the Kāpiti Coast, Wairarapa, Hutt Valley and Wellington. She trained as a physiotherapist in South Africa and has experience in rehabilitation, disability case management and wellness.

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Continued from page 3 ...

Philippa (Pip) McLean joined the Central Region team in October as our Lower North Island-based Community Co-ordinator. Pip has lived in the Hutt Valley all her life and married Ross, 'the boy next door'. They have an adult son, two dalmatians and a cat. Pip is a member of MDANZ, and has lived experience of a neuromuscular condition. She has been involved with the Central branch for several years.

In October, **Melanie Louden** picked up the new role of Communications and Marketing Adviser in the National Support Office. She comes from a background of 17 years as a journalist and editor, and four years in fundraising, media and communications roles at two charities. She has a love of story-telling and is looking forward to meeting members and sharing their stories.

Jane Hazlett joined the Canterbury branch team in November as the Fieldworker for the Upper South Island. She has worked as an occupational therapist for more than 30 years, with the last 22 years in Nelson/Marlborough working in vocational rehabilitation and driving assessment. For five years she managed the Good Companion in-home care service for the elderly and most recently has managed an orthodontic practice.

The Canterbury branch welcomed fieldworker **Ross Paterson** to the team in November. Ross has a social services degree, majoring in career practice and is motivated by working alongside others helping them to find solutions in their lives. He enjoys working with people from all age ranges and walks of life. Ross has worked in the social service area for more than 30 years.

Impact of a ketogenic diet on IBM: A Case Study

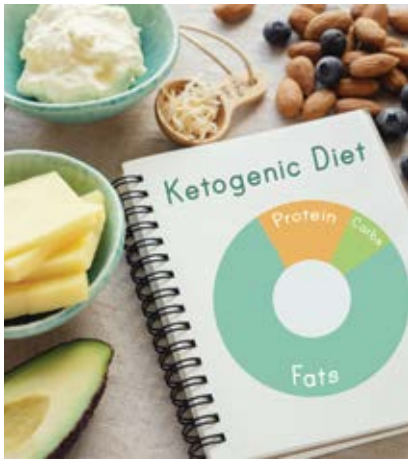
Clinical and research neurologist Dr Matthew Phillips and MDANZ member Pushpa Deo spoke at the IBM Gathering on September 29.

They discussed Pushpa's experience as the first person with Inclusion Body Myositis (to their knowledge) who has used a ketogenic diet as a treatment option.

During his talk at the IBM Gathering, Matt discussed the main problems that occur in the muscles of people with IBM, and how using a ketogenic diet can tackle many of these problems at the same time. Pushpa also spoke about her experience using a ketogenic diet – and how she has been using this as a therapy for more than two years.

The scientific article based on Pushpa's experience was recently published by Matt and colleagues in the open-access journal *Frontiers in Neurology*.

This article provides an overview of IBM and the theoretical benefits that a ketogenic diet could provide. It goes on to describe the experiences of one patient (not identified by name) who used a ketogenic diet as primary therapy. The article describes the improvements in her clinical symptoms and signs, and the stabilisation or positive changes in laboratory tests and radiological (MRI) investigations.



The article can be downloaded in full at the link below:

Phillips MCL, Murtagh DKJ, Ziad F, Johnston SE, Moon BG. Impact of a Ketogenic Diet on Sporadic Inclusion Body Myositis: A Case Study. *Front Neurol.* 2020;11:582402. www.frontiersin.org/articles/10.3389/fneur.2020.582402/full

Matt writes: "It is important to emphasise that this is the first-ever published case involving the implementation of a ketogenic diet in a person with IBM. Thus, although the ketogenic diet holds promise, further studies involving larger numbers of patients are needed before it can be recommended as a proven treatment option."

Matt is a full-time clinical and research neurologist at Waikato Hospital in Hamilton. His foremost passion is to explore the potential feasibility, safety, and efficacy of metabolic therapies, particularly fasting and ketogenic diets, in creating alternate metabolic states that may improve not only the symptoms, but also function and quality of life, for people with Parkinson's, Alzheimer's, Huntington's, and a variety of other difficult disorders.

Adventure and challenges await MDANZ Dukies

Our five young Dukies haven't let the pandemic stop them from continuing to work towards their individual goals, even if it has meant they are taking things a bit more slowly.

Covid-19 lockdowns threw a few spanners in the works for our young members taking part in the Duke of Edinburgh's Hillary Award.

Award Leader Marty Price says five members are taking part in the programme, who "are going very strong" considering the impact of the pandemic.

"It has definitely interfered in what each Dukie has been able to access and this has put them back a bit."

Marty says he is in constant communication with each Dukie and their families to ensure they know he's there to help them when they need it.

"We have changed things around to suit their needs because of Covid-19 and the fact that our Dukies are a high risk in our community, so their health and safety is a priority."

The award is open to anyone aged 14 to 24 years and completing the programme can take as long as the participant wants.

The programme is now run through an app which means it is easy for Marty to keep an eye on each Dukies' progress, and for him to contact them with encouragement and motivation.

The tasks and activities carried out by the award participants are varied.

Jack Lovett-Hurst, 23, from Invercargill and Dylan Schneider, 17, from Auckland both completed their bronze medals last year.

They are now working on their silver medals. Jack has already completed his physical activity – walking.

Ryan O'Rourke, 17, from Gore is busy on his bronze medal. He's already completed his physical activity – pistol shooting and keeping fit, and is learning the acoustic and electric guitar as part of his skills activity and he's also volunteering by taking sports photos.

Nelson-based Joy Gutschlag, 20, is taking part in the bronze programme and has completed swimming for her skills activity and has volunteered as a youth club leader.

Seventeen-year-old Camille Peterson, from Auckland, is working towards her bronze medal and has already completed her voluntary role at a sexuality and gender group. For her skills activity she is training a mobility dog and for her physical activity she is swimming.

For more information go to www.mda.org.nz/What-We-Offer/Duke-of-Edinburghs-Hillary-Award

The sky's the limit for Dylan

Dylan Schneider explains how the Duke of Edinburgh's Hillary Award is opening up new, and impressive, opportunities for him. *By Melanie Louden.*

For Dylan Schneider taking part in the Duke of Edinburgh's Hillary Award is a way to indulge his passion for science and space, while also trying new things.

Since joining the programme in 2017, the 17-year-old Aucklander has completed air rifle shooting and robotics activities, as well as carrying out dog therapy at a rest home to earn his bronze award.

He's now working on his silver award and has sailing and rocketry tasks underway.

The award is open to 14 to 24-year-olds who complete tasks such as physical, skills and voluntary activities, as well as take part in adventurous journeys, to earn bronze, silver and gold medals.

Dylan, who has Duchenne muscular dystrophy, says the programme has been a good way to get him out and about, and experience new things.

So far, the most challenging part of the programme has been carrying out the dog therapy at his nana's

Dylan with his own rocket called a Quarter (Scale) Patriot.

rest home with the family's two greyhounds, Ziggy and Jamie.

Dylan would take his dogs into the rest home and let the residents pat them. "It was difficult to set up – sometimes the dogs didn't want to do things," Dylan says.

However, he says, the residents did look forward to their visits.

Dylan is enjoying the rocketry activities he is currently involved with as a member of the New Zealand Rocketry Association – although it can be a bit nerve-wracking, he says.

"My dad and I go on the first Sunday of every month to the launch site at Taupiri. Lots of people launch their own rockets. Dad and I take our rockets and launch them. When they go up, depending on how powerful they are, you might not see them for awhile and you get a bit nervous."

The teenager got his wish to explore space and science at a greater level when Make-A-Wish New Zealand sent him to Cape Canaveral, in Florida, USA in December 2019.

He "had a great time" watching two rocket launches and, of course, visiting Disneyland and Universal Studios.

Dylan says he has enjoyed all the new opportunities that have come his way through the Duke of Edinburgh programme and encourages anyone looking to try new things to get involved.

Dylan at the Kennedy Space Centre, Cape Canaveral, Florida, USA, watching the Spacex Falcon 9 Rocket Launch.

Dates for your diary

The MDANZ National Support Office will close for the Christmas holidays on Friday, December 18. The office will open again on Tuesday, January 5.

A cup of tea and a catch up with Talitha Vial

Each issue we introduce a MDANZ team member.

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

I have worked as a Wellington Central Fieldworker since 2019. However, I joined MDANZ 25 years ago as a member.

What qualifies as a great day at work for you?

When I have had a good conversation with the members I meet or have been able to make a difference in someone's life.

On a private note, a good day for me is if I don't have to collapse into bed at 7pm because I have used all my energy that day and am feeling weak. A great day is not having fatigue that day.

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

I would love to see each and every one of our members enjoy achieving something they would love to do, whether it is feeling sand and sea between their toes; or looking at the clouds below them; going on a date or just having a cuppa with someone special.

What are you passionate about?

I am passionate about a lot of things – I would probably say I'm passionate about life.

I know how delicate our life can be so I tend to enjoy the small things a bit more than usual.

I really enjoy spending time with, or chatting to, people with disabilities.

But I have a bias towards Myasthenia Gravis, which I have had for 25 years, and I have also developed a passion for disability rights.

But to answer the question... people... I'm passionate about people.

CanIPlayThat.com

A website that is created, managed, and written by disabled gamers, reviews games and hardware with a focus on accessibility for all sorts of disabilities. *By Scott Boyle.*

Christmas is fast approaching and hopefully your gift shopping is well under way. 2020 has been a rather turbulent time for Kiwis, and the world in general, but due to lockdowns many people have become more accepting of a rather immersive hobby – gaming.

Chances are we all know kids, teenagers and adults that happily pass the time by playing video games, or perhaps you even enjoy a bit of gaming yourself, whether in multiplayer titles with friends, or solo in narrative-centric games.

As the gaming industry becomes more and more popular, we're seeing some of the greatest stories being told

in the medium, along with critically acclaimed writers, actors, directors, and composers working within it. There are even professional gamers and streamers making millions of dollars throughout their careers and raising awareness for worthy causes.

But what about disabled gamers?

For many of us that have physical struggles it can sometimes be difficult to play certain games or use various controllers and setups. Fortunately, there is an incredible website that is created, managed, and written by disabled gamers. These amazing people review games and hardware with a focus on accessibility for all sorts of disabilities. They even work alongside major development studios to help ensure accessible options are included and optimised.

So, if any of our amazing members and their families are considering purchasing a new game, or one of the newly released consoles, for Christmas, take a peek at CanIPlayThat.com for ideas about what works best for you.

Scott Boyle is a member of the MDANZ National Council.

Empowering Kiwis with disabilities

Eden Park and The Cookie Project have announced a partnership which sees the social enterprise operating from a kitchen within New Zealand's national stadium, creating more employment opportunities for Kiwis with disabilities.

Founded in 2018, The Cookie Project provides meaningful employment to Kiwis with disabilities by hand-making cookies and paying at least the adult minimum wage of $18.90 an hour. Since its inception, more than 2,400 hours of

employment has been generated.

Co-founder and CEO at The Cookie Project, Eric Chuah, says in a statement that the impact of the first lockdown left the social enterprise without access to safe premises for its employees to bake from.

He says that after reaching out on social media for urgent assistance, the team at Eden Park connected with the project and within 10 days they had a new home.

"We're incredibly thankful for the lifeline they've given us and our bakers – we're now gearing up to prepare for the festive trading season with our 2020 Christmas Cookie Bundle. This wouldn't have been possible without Eden Park's support to create a more diverse and inclusive Aotearoa," Eric says.

Eden Park CEO Nick Sautner says the opportunity to welcome The Cookie Project to the stadium aligns with the organisation's ongoing community values. "The organisation is committed to being an inclusive venue for Auckland and New Zealand and community is at the core of our Game Plan".

See www.thecookieprojectnz.com for more information.

Do you have LGMD?

The NZ NMD Registry has been approached about a possible project to help recruit for an upcoming LGMD study.

The New Zealand Neuromuscular Disease Registry is part of the TREAT NMD network of neuromuscular disease registries. It has recently been approached about a possible project to assist with recruitment for an upcoming limb girdle muscular dystrophy study involving people with LGMD types 2C, 2D, 2E. These forms of LGMD are all sarcoglycanopathies where damage in one of the four genes that make up the sarcoglycan protein complex means the protein complex is unstable and results in limb girdle muscular dystrophy.

The NZ NMD Registry responded to the enquiry with information about the number of Registry participants with these types of LGMD. The Registry was also asked whether NZ provides genetic screening for LGMD, which it does. However, it is very aware of a backlog of undiagnosed LGMD patients.

It was also asked whether the Registry includes LGMD patients without a confirmed genetic diagnosis, which it does with 30 plus people enrolled with an unconfirmed clinical-only diagnosis.

One of the aims of the Registry is to help these people to receive confirmation of their diagnosis. This is particularly important when some neuromuscular conditions that look like LGMD, such as Pompe disease or congenital myasthenic syndrome, have treatments readily available. The upcoming LGMD study, mentioned above, will rely on participants having a confirmed genetic diagnosis of LGMD 2C, 2D, or 2E. So, if you have LGMD but don't know what type, you need to: (1) Request your GP refer you to neurology. And then ask a neurologist to request that a gene panel is ordered. (2) Enrol with the NZ NMD Registry – it can also help with number 1.

The Registry looks forward to learning and sharing more about the study with the LGMD community. Registration and consent forms are on: www.mda.org.nz/Our-Research/NZ-NMD-Registry. Or contact your fieldworker, or the Registry direct on nznmdregistry@adhb.govt.nz.

Smiling into the storm

When the world throws a year of chaos at me, how will I smile in the storm?

In an open letter to MDANZ members, David Sanders, a well-being manager at EAPworks, looks at how to cope with life when it's difficult, or just plain overwhelming. He offers tips on how to be positive, see beauty in the mess and encourages us to be the person who breathes hope and laughter in the midst of it all.

Hi everyone,

Well, I don't know about you, but this year has certainly confronted me and my friends with a life we never expected.

Over the last year or two, we have been so caught up worrying about the big issues in our world, like global warming, terrorist attacks, earthquakes, volcanoes, the economy and more, that none of us expected our world to be derailed so powerfully, by an insignificant, tiny, unseen virus.

The fallout of this virus, (known as Covid-19) has impacted us in multiple areas of our lives, from work issues, to personal relationships, to finances, mortgages, rent, buying groceries, self-care, personal hygiene and, of course, forced isolation.

The impact of lockdown in itself, has also compounded many of these issues leaving many of you feeling vulnerable, alone and, at times, frightened.

These are just a few of the issues forced upon us all and I just want to acknowledge them. I know my list isn't exhaustive and for many of you reading this, it doesn't even begin to cover all the stressors you've encountered, both minimal and huge, perhaps even life threatening.

It's been hugely challenging for some and downright frustrating and exhausting for others.

Many of us would consider ourselves robust or resilient at the best of times. People who don't easily buckle under strain and yet, because of the number of issues that have been washing over us continually for eight months, we have found our resilience challenged, our coping skills questioned and our vulnerabilities exposed as we have battled with exhaustion, uncertainties, and other anxieties and fears.

So, if you're reading this today feeling like you are "over this" and wondering when it will all cease, then I sympathise with you.

You are normal. You are allowed to feel tired and overwhelmed from it all. Why? Because we all are. We have been facing unprecedented stressors not seen before in our country since the last great war.

Rachel Remen, author and teacher of integrative medicine, once said: "The expectation that we can be immersed in suffering and loss daily and not be touched by it is as unrealistic as expecting to be able to walk through the water without getting wet."

How true that is. However, it doesn't really solve our problem does it?

How to cope with life when it's difficult, or just plain overwhelming? How to be positive, see beauty in the mess and feel positive about our situations?

I hope this sounds respectful, as I don't mean to take away from the seriousness of your lives. However, in my work as a health worker, I've realised certain things.

1. We either let our circumstances dictate to us, and our well-being, or we dictate back to our circumstances.
2. We become what we focus on.
3. Only I can breathe life into my day. It's all about my focus, my attitudes and my coping skills.
4. When the world throws a year of chaos at me, how will I smile in the storm? What will be my focus and how will that focus allow me to laugh and smile and see beauty in the middle of my difficulties? This is my challenge.

So here are a few tips to help you smile in the storm and re-centre your focus over Christmas and well, who knows how long? These aren't rocket science, but they are powerful and true.

Gratefulness, appreciation and laughter

Pain always demands to be noticed and unless you challenge its "talk", you will focus in on it. You will become what you focus on! You may even find yourself buying into its anxieties and fears. So… choose who you want to be for you and your family.

I encourage you to be the person who breathes hope and laughter in the midst of pain.

So on that note, I'm going to encourage you to redirect your focus to **gratefulness, appreciation and laughter**.

You are allowed to feel tired and overwhelmed from it all. Why? Because we all are. We have been facing unprecedented stressors not seen before in our country since the last great war.

These three things have huge medical and psychological benefits during times of stress.

They release wonderful endorphins that build disease-fighting chemicals and hormones such as DHEA. It's a natural pain killer and reduces cellular decay – it releases tension and helps to build strong immune systems and changes the way we see our environment. See www.askthescientists.com/laughter-immunity/ and www.usa.edu/blog/how-laughter-can-relieve-stress/

Do you all remember Dr Patch Adams? He was so convinced of its impact medically and psychologically, that he devoted his hospital to encouraging everyone, (especially staff) to practice these three important tools for health.

Psychologically, the three of them build resilience, take away the power of our anxieties and soothe our fears.

They change our perspective and make us more attractive to others. They help our children and grandchildren to view pain without fear. Now that's huge.

"There is nothing in the world so irresistibly contagious as laughter and good humour." – Charles Dickens.

"I have seen what a laugh can do. It can transform almost unbearable tears into something bearable, even hopeful." – Bob Hope.

Self-care

Over 36 years in my work I've discovered that every human being is unique in discovering what breathes life into them during stressful times and it's important to embrace

Panic breeds panic, but a smile and... a reassurance breeds safety. Rob the situation of its potential power.

your particular way of re-invigorating yourself.

So, take time right now to stop and ask yourself: "What re-energises me? What breathes life into me? What allows me to smile in the storm and say, I will be okay because..."

Here's a good site to go to, that gives lots of self-care tips www.psychologytoday.com/nz/blog/click-here-happiness/201812/self-care-12-ways-take-better-care-yourself.

However, if you are lost for ideas, here's a few to get you started. Don't ignore the power of breathing life into your day.

- Talk with a positive trusted friend.
- Exercise.
- Eat the right foods.
- Engage in a hobby.
- Look after your spiritual and cultural needs.
- Debrief/offload with a caregiver, professional.
- Watch a funny video.
- Write, sing, create, explore, pray, laugh, love life.

Managing anxiety for families

And lastly, I've had a lot of requests from families wanting tips on how to manage anxiety for themselves and their children. There is no way I can fit all this in here, but here are some simple tips to help bring those anxious thoughts under control.

- Daily routine is important in calming through change. Keep it up.
- Limit social media at night and unnecessary info during the day.
- Family culture is paramount. Reinforce it regularly.
- (You become what you focus on.) Channel the focus.
- Use stories from your own life as examples, rather than information, facts and lectures.
- Always place "okay-ness" into your worries and anxieties. Model courage.
- Use the Covid situation to engage in laughter and fun. (Remember: Either your circumstances will dictate to you, or you will dictate to your circumstances.)
- Keep your emotional reactions to unforeseen situations calm. Use a reassuring and safe voice. Panic breeds panic, but a smile and an okay reassurance breeds safety. Rob the situation of its potential power.
- Don't verbalise your fears, rather verbalise that you're the master of those fears and we will be okay.
- Set expectations and boundaries on attitudes, hysteria, devices, respect and kindness.
- Encourage family members to express their fears and questions. Just don't react to them. Show confidence and support. Teach them to find their strength in the storm.
- Don't be a helicopter parent that rescues. Rescuers create victims and victims get angry. Be a helper, that teaches coping skills.

It's been a pleasure chatting to you all. Merry Christmas.

David is an EAPworks Well-Being Manager. He has been with EAPworks for 18 months and has 34-years experience in the mental health field. His background and specialities are in terminal illness, depression, suicide, critical incident and mental well-being. David lives in a little settlement called Puhoi, just north of Auckland. He has three adult children and in his spare time paints, loves art and fishing.

Need to talk?

Through your MDANZ membership you and your family can have free, confidential counselling to help address a myriad of life issues.

Our mental health, wellbeing and life skills are a vital part of coping with everything that life throws at us. And 2020 has thrown a lot at us all. But sometimes we don't know who to call on, or who to reach out to.

Did you know that MDANZ offers its members, and those close to them, free and confidential counselling through EAPworks? EAPworks is a network of NZ practitioners, counsellors, psychologists and other providers and their services are available for you to use. Their team can assist with things like stress; work issues; relationship or family issues; grief and loss; drug and alcohol issues; anger or conflict issues; life transition and direction; health and wellbeing; bullying and harassment; career planning; help with your CV; budget assistance; depression and anxiety. MDANZ funds up to three counselling sessions per person per year. EAPworks can request funding for further sessions and these will be considered by MDANZ on a case-by-case basis, depending on the need and available funds.

For more information go to www.mda.org.nz/What-We-Offer/Counselling-Service.

The service is strictly confidential – at no point does MDANZ know who has reached out to EAPworks. You will have 24-hour, 7-day-a-week access to personal telephone support. For more information about EAPworks go to www.eapworks.co.nz , phone 0800 735 343 or talk to your fieldworker.

There are plenty of other options out there when you, or someone you know, needs help with mental health.

- **1737** www.1737.org.nz. Free call or text 1737, 24 hours a day to talk to, or text with, a trained counsellor or peer support worker.
- **The Mental Health Foundation** www.mentalhealth.org.nz. The website has information on mental health conditions, where to get help and how to support those you love and care about.
- **Depression.org.nz** www.depression.org.nz, 0800 111 757 or text 4202. Talk to a trained counsellor about how you are feeling or to ask a question.
- **Lifeline** www.lifeline.org.nz, 0800 543 354 (0800 LIFELINE) or free text 4357 (HELP). Call or text for 24/7, confidential support from qualified counsellors and trained volunteers.
- **Suicide Crisis Helpline** www.lifeline.org.nz/services/suicide-crisis-helpline, 0508 828 865 (0508 TAUTOKO). A free service available 24 hours a day, 7 days a week operated by highly trained and experienced telephone counsellors who have undergone advanced suicide prevention training.
- **Samaritans** www.samaritans.org.nz, 0800 726 666. Confidential, non-judgemental and non-religious support.
- **Mentemia** www.mentemia.com. This app coaches mental wellbeing and is free to all New Zealanders - ideas and tools to help you learn how to be well and stay well. It deals with the most common stressors - poor sleep, anxiety and stress.

For the young

- **Youthline** www.youthline.co.nz, free call 0800 376 633, free text 234, email talk@youthline.co.nz or web chat from 7pm–10pm www.youthline.co.nz/web-chat-counselling. Counselling by phone, text, chat, Skype and in person. They also offer mentoring and employment goals and advice.
- **The Lowdown** www.thelowdown.co.nz, email team@thelowdown.co.nz or free text 5626 . The website helps young New Zealanders recognise and understand depression or anxiety.
- **What's Up** www.whatsup.co.nz, 0800 942 8787. A free, counselling helpline and webchat service for children and teenagers. Phone counselling - Monday to Friday, noon–11pm and weekends 3pm–11pm. Online chat - Monday to Friday from 1pm–10pm and weekends 3pm–10pm.
- **Kidsline** www.kidsline.org.nz, 0800 54 37 54 (0800 kidsline). A 24/7 helpline for children and young people, run by specially trained youth volunteers.

Coping with the loss of a son and brother during lockdown

Doctors, nurses, counsellors and their strong faith support this tight-knit family.

The Level 4 lockdown brought about by the Covid-19 pandemic was a sad but also special time for the Taylor family. *By Melanie Louden.*

Sharyn and Steve Taylor say the Level 4 lockdown restrictions that were in place when their son Hamish passed away in April, meant their family was given the chance to slow down and be with him.

"It was like God made it this way so we could cope with the loss of our son," Sharyn tells *In Touch*.

Hamish and his younger brother Austin were diagnosed at birth with Duchenne muscular dystrophy (DMD). Hamish was in a wheelchair by the age of 10 and needed full-time care to bathe and dress by the time he was 13. By 15 he could no longer feed himself.

At the beginning of April, Hamish, 23, was "not feeling like himself". He was sick for the whole month and the family worked with St John, the local hospice and their GP to develop a plan to help with his pain, anxiety and to keep him comfortable.

"During the last two weeks, he struggled to spend time in his wheelchair and by the Thursday before ANZAC Day, he was not able to be in his chair anymore," Steve says.

"Our parish priest visited on the Friday night and we supported Hamish in our family bubble at his bedside until he passed on the Sunday morning."

Hamish died on the morning of April 26, at his Hamilton home, surrounded by his family – Sharyn, Steve, Austin, 21, sister Lucy 14, and Floyd the family dog.

Steve says the Level 4 lockdown was a special time for the family because they were able to dedicate their time to Hamish.

L to R: Hamish with his sister Lucy, and brother Austin.

The Level 4 lockdown was a special time for the family because they were able to dedicate their time to Hamish.

"We spent that last month with him, without having to rush off to work or school," Steve told the *Waikato Times* in an article in May. "Although we couldn't get our Taranaki family up to visit, it was just us. We were able to be with him."

He says the family had "time to ourselves to spend with Hamish, and handle the emotion ourselves".

Sharyn says she always worried about having people coming in and out of the house and that would have been overwhelming for her.

"When he did pass, it was like a sense of peace and he went straight into God's embrace."

Sharyn's sister, Hamish's aunt, Denise Baker lives in Taranaki and says not being able to see Hamish before he died and be with her sister's family "was the hardest thing I have been through".

"I even wrote an email to Prime Minister Jacinda Ardern asking her to let us go to Hamilton! I was grateful that Sharyn and Steve had the support of each other during this time."

To add to the family's grief, Sharyn, Denise and their brother Mark's Nana passed away the week before Hamish died.

"We are a tight knit family and it was hard to have the barrier of the Covid restrictions," Denise says.

But the Taylors weren't going to let Hamish's death pass without celebrating his life – once lockdown levels allowed it, family and friends came together at their parish on July 4.

"Hamish was in the bridal party when his cousin Crystal married on that date in 2015 and it is also American Independence Day. Hamish was fascinated with the USA," Steve says.

He says having the memorial so long after Hamish's death turned out to be a blessing.

"It gave us time to plan the memorial the way we wanted it. We were able to take our time.

"It was more of a celebration because we had gone through the grieving stage."

The family say the support they received from their parish priest, their GP, and the "tremendous" support from Waikato Hospice nurses and counsellors, helped them find the resilience and strength to get through such a difficult time.

"We just supported each other the best we could."

Throughout lockdown, the Taylors still received the same personal care support, but Austin missed out on his cooking class.

The family passed the time by walking Floyd, watching *Amazing Race* episodes and building lots of Lego. Lucy found going to the supermarket to be a highlight.

Hamish with the family dog, Floyd.

Austin and Lucy say Hamish was a great big brother who always looked out for them while Steve and Sharyn proudly point to their "very kind-hearted and caring" son.

"He had a serious side and had opinions on world matters, especially Mr Trump!"

California Dreaming

In Touch first met Hamish and his family in Winter 2019 when he told our readers about fulfilling his dream of visiting America to celebrate his 21st birthday.

Steve, Sharyn, Hamish and Lucy visited San Francisco, Los Angeles, Las Vegas and the Grand Canyon.

Lighting up the world

The magic of Christmas from Aaron's computer

Aaron Collins has been bringing joy to his Palmerston North community for the past 15 years with huge synchronised Christmas light and music displays outside his home. And he's not letting the pandemic stop him from doing the same thing this year. *By Melanie Louden.*

Aaron Collins' fascination with technology, cables, wires, computers and all things electrical began early.

As a youngster he was delighted to receive an extension cord for his fifth birthday, but it was a trip to visit an uncle in Australia as a nine-year-old that sparked his love of Christmas lights.

Aaron's uncle had a few lights up around his house, and the young boy was fascinated by them, so much so that when the family got home, he insisted they have lights of their own.

"We put up a few fairy lights," says Aaron's mum Anela Collins. "I wanted them in the back yard so I could see them, but Aaron wanted them out the front."

The rest, as they say, is history.

Aaron, 31, has Duchenne muscular dystrophy (DMD), and has been on a ventilator since he was 16 years old.

As a young adult, Aaron started buying his own lights and "the collection just built up".

He's now been creating large Christmas light displays outside his home for his Palmerston North community to enjoy for the past 15 years.

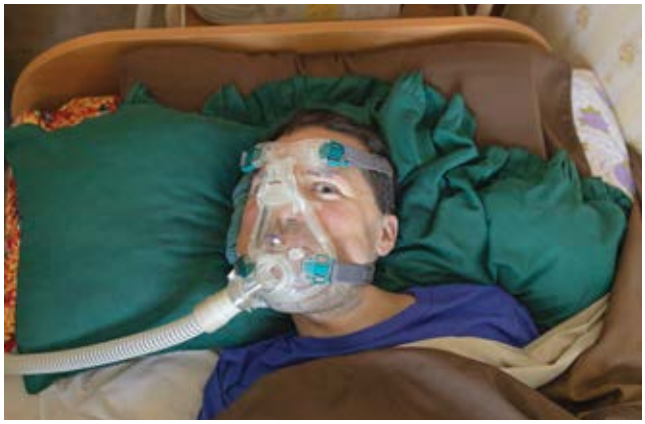
He coordinates the display of synchronised lights and music via computer from his bed, while a small team of helpers, including Anela, executes his vision. A neighbour provides the speakers and wires them up.

"It used to take a weekend to put the lights up. Now it takes three months. We start in September," Anela says.

Aaron spends six months of the year on the computer sequencing the display, buying new lights and organising his helpers.

They've never counted how many people have visited their home each festive season, but Aaron guesses it would be a "couple of hundred".

While Aaron can't join the revellers very often, when he is able to get up, he enjoys seeing the joy the lights bring to people. Otherwise Anela goes outside with her phone and Skypes Aaron so he can still see people's reactions.



Aaron with his parents, Anela and Graeme and brother, Nathan. Photo credit: James Collinson.

Aaron spends six months of the year on the computer sequencing the display, buying new lights and organising his helpers.

"People come in and laugh and smile, and it makes Aaron feel good," Anela says. "It's worth all the hard work."

She says her son definitely has a love of technology and computers and putting these displays together is "a challenge for him".

He sources most of his lights from America, buying them in January when they are on sale.

Aaron's collection now stands at 150 sets of lights, and his favourite display is the mega tree – a cone shaped display that initially looked like a tepee until he bought a star to go on the top.

Aaron's condition was picked up when he was three-years-old after one of his kindergarten teachers pointed out to Anela and her husband Graeme that the youngster avoided stairs, and had trouble getting up off the floor.

Doctors' appointments and testing confirmed Aaron had DMD. Testing also confirmed his younger brother Nathan had the condition.

Well prepared for Covid-19

The Collins family say they were mentally well prepared for the impact Covid-19 was likely to have on society, but it didn't make adjusting to lockdown any easier.

"Nathan has a degree in microbiology, so he knew from the early reports in December that the virus was going to be serious," Anela says. "He didn't cause us to panic, but his knowledge of how viruses work was helpful."

She says they were anxious as the country went into lockdown.

"It was quite a rush to get organised especially with our caregivers. It was hard to get supplies of hand sanitiser and masks."

While Aaron can't join the revellers very often, when he is able to get up he enjoys seeing the joy the lights bring to people.

The family normally has nine caregivers, but one of them went back to India and another was stood down because she also worked at the hospital.

"Even though we were down two caregivers everybody helped out and we got through it really well."

Another stressful task was getting groceries – at one point Aaron stayed up until 1a.m. to get a delivery slot.

"We felt it was too risky to go to the supermarket."

During lockdown the Collins' were also coping with Graeme's mother's struggle with cancer, and her passing in August.

But amongst the anxiety and sadness, there were positives for the family – there were no appointments to rush to, Graeme was home all the time and could help more with the boys, it was a restful time for them, and they could attend church online.

For now they are simply enjoying sharing Christmas with their community in the brightest way possible.

Lunch at Te Māhia on Kenepuru Sound.

The road to recovery

Michelle's road trip

Dr Michelle Smith isn't letting Covid-19 get in the way of her holiday plans. Michelle and her husband Blair switched the Mediterranean for the South Island and learnt that there are plenty of accessible options open to travellers in New Zealand who are willing to ask the right questions.

While NZ is still feeling the impact of Covid-19, at least we can move around freely, although with caution and increased hygiene as our travelling companions.

Domestic tourism is our road to recovery. Taking a break and seeing our beautiful backyard is good for our economy and for the soul. It helps us heal emotionally, socially and physically.

As I write this, my husband Blair and I should be cruising the Mediterranean – a bucket-list trip we had been planning and saving for, for years.

As Covid scuppered that plan, we decided to explore the South Island. Between late October and early November, we spent a wonderful three weeks traversing 4000km, absorbing breath-taking scenery; enjoying wonderful experiences; catching up with family and friends; eating fabulous food and staying in some lovely accommodation.

... we spent a wonderful three weeks traversing 4000km, absorbing breath-taking scenery; enjoying wonderful experiences...

I love planning trips and, as usual, I did a lot of research into accessible activities and accommodations. We took our own vehicle, so that was one less thing to have to consider.

We travelled from Hawke's Bay to Wellington where we stayed the night before catching an early ferry to Picton. First time on the ferry, and we were most impressed

with the Interislander from booking to check-in to overall access onboard. The Kaitaki is a newer vessel and the most accessible of the company's three ferries.

Our road trip saw us stop in Blenheim; Geraldine; Tekapo; Wanaka; Queenstown; Te Anau; Oamaru; Ohoka and Picton.

Wheelchair accessibility is important to us, and many questions are asked of accommodation providers. The perfect accessible accommodation is hard to find, but we hit the mark pretty close on most of our motels, hotels and B&Bs.

We stayed in a couple of historic properties – being a trained historian, old buildings fascinate me and when they are accessible, I just have to stay in them!

Even when not fully accessible (i.e. plastic outdoor chair in the shower rather than a fixed seat), such as the Old School north of Oamaru, by asking the right questions of the owners we knew we could manage with a little more effort.

I was determined to stay in this lovingly restored historic building, and we were not disappointed.

In fact, the owners asked my advice on the type of seating for the shower – due to the size of the bathroom and the configuration of the wet-area space it could not take a fixed seat - and ordered a height-adjustable shower chair while we were there!

Wineries, museums, historic settlements, river walks, snow-capped mountains, vast oceans, seals, penguins, blue lakes, steamships, and gushing waterfalls only hint at the things we saw and did.

We ate at some great cafes and restaurants – Rata in Queenstown and Nin's Bin on the Kaikoura coast were particular favourites.

We took the scenic routes - highways 70, 72 and 8 - to get a sense of mainland New Zealand. Vast valleys and towering mountains gave way to lush farmland before changing to rocky outcrops and heather clad hills.

So many highlights, such as the stunning beauty at the Church of the Good Shepherd at Tekapo, the majesty of Milford Sound and the little Blue Penguins of Oamaru who sat at my feet, but sadly there is not enough space to write about them here.

However, I am happy to be contacted via the MDANZ if anyone would like further information, recommendations, or our full itinerary.

Top: Blair and Michelle at Milford Sound. *Above:* Michelle at Mirror Lakes on the road to/from Milford Sound.

We were most impressed by the willingness of accommodation hosts, tour operators and the general public to assist in making things work for us, particularly when accessibility was limited.

The Dark Sky Crater Experience in Tekapo and the cruise on Milford Sound are two such examples whereby with some forward thinking and burly guides/employees to assist, experiences usually inaccessible became very much do-able. Barriers can be broken down and often there is an adventure to be had in doing so.

Even when accessibility was good, the friendliness and exceptional customer service we received made every experience memorable.

We do live in a stunningly beautiful place and I encourage you to get out and explore if, and when, you can.

** Dr Michelle Smith is the Community Coordinator for the East Coast District of MDANZ's Central Region and has facioscapulohumeral muscular dystrophy.*

Flynn and his family are planning more outdoor biking adventures over the summer.

Flynn's wheels bring freedom and independence

New opportunities for a very sporty teen

A "spectacular" recumbent bike is seeing 17-year-old Flynn Mitchell get back to the outdoors again, which is having a positive impact on his mental well-being and… is leaving the rest of his family in his dust. *By Melanie Louden.*

A positive mindset, the willingness to take risks, and some vital support from the Bradley Jenkin Memorial Fund is seeing Flynn Mitchell make the most of the outdoors again.

Flynn has Friedreich's ataxia and uses a mobility scooter to get around, and a recumbent bike, which he works with his legs, to keep up with his sport-loving family.

The "spectacular" recumbent bike, which cost approximately $16,300, was bought with the help of fundraising by friends, and a $1,000 grant from the Bradley Jenkin Memorial Fund.

Flynn says his new wheels give him freedom, independence and the opportunity to have fun, while hopefully slowing down progression until a cure for Friedreich's ataxia is found.

Flynn is very grateful for having "such great friends" and financial assistance from the fund.

The 17-year-old, from Auckland, says getting back outdoors has had a positive impact on his mental wellbeing.

"Pick something you know you will use, and which will bring joy and be worth the purchase. Try before you buy."

"It was important for me to get a recumbent bike so that I could get out and about with my family again, and do some outdoor sport in my free time that is not at the gym.

"After years of struggling to keep up with the rest of the family they now 'smell my dust' trying to follow me."

Flynn is very grateful for having "such great friends" and financial assistance from the fund.

"Without their help and support I might not have been able to get the bike which means freedom and independence for me."

While Flynn waited for his recumbent bike to arrive, he had a rental which he has taken to Muriwai, the Hunua Ranges, Waitawa Regional Park and he has done part of the Hauraki Rail Trail.

The plans don't stop there. Flynn, his parents Scott and Gesa, and his brothers Peer, 15, and Yonas, 13, are planning on cycling the Central Otago Rail Trail over the Christmas holidays and there are plans to complete other trails throughout New Zealand during summer.

Flynn was diagnosed with hypertrophic cardiomyopathy "out of the blue" in 2014. The disease sees the heart muscle become abnormally thick, making it harder for the heart to pump blood.

"I also started getting some spinal and balance issues and people around me realised that there must be something else going on," Flynn says.

"I went from specialist to specialist and had a genetic test in 2018 which confirmed the diagnosis of Friedreich's ataxia."

The sport-loving teenager says the progression of the condition meant he was becoming quite isolated, he could no longer walk around school, and had to adapt the sports he plays and how he takes part in them.

"My condition has progressed in the way that I now have to make key changes in my life, such as [using] different methods of mobility and different ways that I go about things, like using a recumbent bicycle.

"I used to walk around school by myself, played sports and went hiking. Now I stick to the gym and use a mobility scooter. I am currently involved in rowing as a coxswain and training at the pool, and I've picked up adaptive skiing, which I really enjoy."

Flynn has even managed to combine his love of sports with fundraising for Friedreich's ataxia.

In May he took part in the "Lend Me Some Muscles" campaign, raising $4,000 for the Friedreich's Ataxia Research Alliance (FARA).

In October a team of New Zealand fundraisers, organised by Flynn and his family, took part in the Global Ride Ataxia Challenge and raised $18,971 – some $3,900 more than their initial goal, and contributing to the US$444,921 raised globally for FARA.

The money will go towards research to find a cure. Flynn says he wanted the challenge to be held in New Zealand to raise awareness about the different challenges people face.

Flynn is encouraging MDANZ members to make use of the Bradley Jenkin Memorial Fund.

"Pick something you know you will use, and which will bring joy and be worth the purchase. Try before you buy. We hired a bike for six months and I feel I have made a very informed decision and it's what I really wanted."

The Bradley Jenkin Memorial Fund

The Bradley Jenkin Memorial Fund is designed to help 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 fees and course fees.

For more information about the Bradley Jenkin Memorial Fund, go to https://www.mda.org.nz/What-We-Offer/Bradley-Jenkin-Memorial-Fund

Metabolic Myopathies

They are rare, highly variable in how they present, and often misdiagnosed early in the disease course - but few conditions rival the interest that metabolic myopathies elicit, writes Shanthi Ameratunga.

Enthusiastic experts across the breadth of health disciplines are researching these conditions - from molecular geneticists, basic and laboratory scientists, to a range of medical specialists and allied health professionals.

For many people diagnosed with these conditions, the rapidly evolving scientific knowledge on the causes, consequences and treatment options, could not come soon enough.

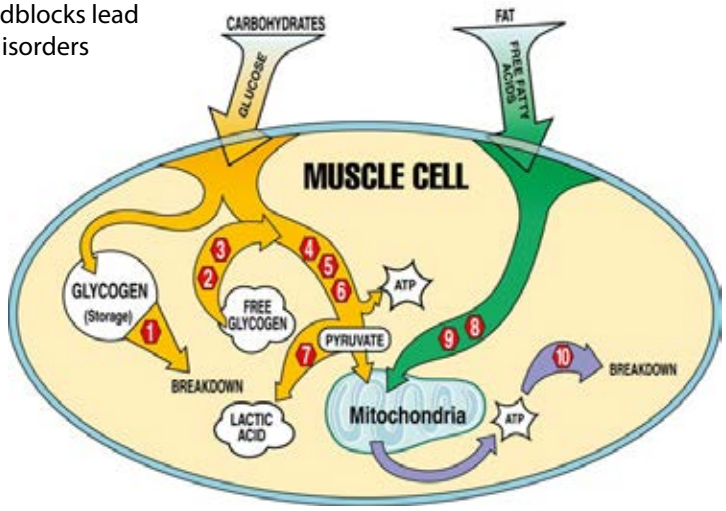
This brief overview describes the key features of this curious and complex group, with a summary of one the best-known conditions, Pompe disease.

Metabolic myopathies are a family of inherited conditions caused by errors in gene or DNA codes, where particular enzyme defects act as roadblocks in biochemical pathways that store, release or use body fuels (hence the term metabolic).

These defects lead to variable degrees of muscle impairments (hence the term myopathies).

The fuel pipelines affected are those metabolising glycogen (stored form of glucose), fatty acids, or purine. Some related conditions interrupt the transport of energy units into or out of mitochondria (energy-pumping engines in cells).

Roadblocks lead to disorders



The Figure, used with permission from Muscular Dystrophy Association, Inc. shows the connections between enzyme defects in some of the many metabolic myopathies.

Roadblock or deficiency linked to numbers in the Figure *(above)*	Disease name
Glycogen storage disorders	
1. Acid maltase or Acid alpha glucosidase (GAA)	Pompe disease, Glycogenosis Type II
2. Muscle phosphorylase	McArdle disease, Glycogenosis Type V
3. Debrancher enzyme	Cori or Forbes' disease, Glycogenosis Type III
4. Phosphyfructokinase	Tarui disease, Glycogenosis Type VII
5. Phosphoglycerate kinase	Glycogenosis Type IX
6. Phosphoglycerate mutase	Glycogenosis Type X
7. Lactate dehydrogenase	Glycogenosis Type XI
Fatty acid oxidation disorders	
8. Carnitine palmityl transferase	Carnitine palmityl transferase deficiency
9. Carnitine (amino acid that helps transport long-chain fatty acyl co-enzyme A)	Carnitine deficiency
Purine metabolism disorders	
10. Adenosine Monophosphate Deaminase	AMPD deficiency

Some of the common features

Each condition is unique and the way they present in different people can be highly variable. But there are some features that occur commonly.

Genetics: Most of these conditions are inherited in a pattern referred to as autosomal recessive. That is, a copy of the abnormal gene code is passed down by both parents. This is no one's fault, these conditions are not due to anything that was done, or not done, during pregnancy. Too often, families carry a lot of guilt or shame, made worse by the lack of awareness and misinformation in wider society. Clinical genetics services play an important role here, undertaking the necessary tests and supporting families and health professionals with the best available evidence and guidance.

Clinical presentations: In general, problems arise from reduced energy levels or impaired functions of some systems when fuels that are not getting metabolised accumulate in higher levels in body organs (e.g., glycogen storage disorders).

The age when the disease becomes apparent and its severity often depends on the amount of active enzyme in the body – and this can vary even in the same family.

The common symptoms are muscle pain, cramps and weakness; and exercise intolerance and fatigue. In some forms of these diseases, dysfunction of heart and respiratory muscles will shorten lives. Some conditions also lead to brain and endocrine dysfunction, and acute (or sudden) destruction of muscle cells, sometimes precipitated by infections.

At the other end of the spectrum, there are disease forms that are far less troublesome and compatible with normal life spans.

Diagnosis: Experts acknowledge that even in countries with access to high quality specialty services, the number of people investigated and diagnosed under-estimates the real occurrence of metabolic myopathies in the population. There is no simple playbook to identifying these diseases. The symptoms and signs are non-specific, occurring more often in other conditions, many of which are more common and resolve spontaneously.

Unless a family member has been diagnosed already, the penny may drop only when symptoms get more complicated. With the power of hindsight, people recognise tell-tale symptoms that have lasted years. So diagnosing these conditions requires a high degree of clinical suspicion and detective work. Following a thorough clinical assessment, investigations start with blood tests. More specific muscle biopsies and physiological tests are increasingly giving way to next-generation gene panel screening for several metabolic myopathies. In New Zealand, these are coordinated by specialist services.

Treatment: Most patients are assessed by multi-disciplinary teams that arrange supportive care including exercise and nutritional interventions that maintain energy levels, physical therapy, respiratory care, and advice on how to reduce risks that can trigger disease deterioration. Enzyme replacement therapy has heralded a step-change in progress for Pompe disease and novel molecular approaches hold promise as definitive treatment options on the horizon.

Not discounting the huge need and value of discovering life-changing treatment options, I was struck by a notable absence when reviewing the literature. That is the relative lack of information on interventions that could address structural barriers (e.g., in education, employment, housing) that are also likely to have profound impacts on the social and economic opportunities of people with metabolic myopathies.

By providing voice and power to people with lived experience, MDANZ is on an important mission in this regard.

Professor Shanthi Ameratunga is MDANZ's Clinical and Scientific Advisor.

Pompe disease

First described in 1932 by J.C. Pompe (a Dutch pathologist), this autosomal recessive disease, also known as Glycogen Storage Disease type 2, is caused by mutations in the GAA gene, which codes for alpha-glucosidase (also known as acid maltase). The reduction in the effective enzyme inhibits the conversion of stored glycogen to free glucose which serves as energy fuel for cellular function.

In the congenital or infant forms, the GAA enzyme is markedly reduced or near absent, and without treatment, progressive muscle weakness, breathing difficulties and heart involvement shortens life spans. In the late (juvenile or adult) onset forms of the disease, the enzyme deficiency is less marked, symptoms are milder, and people live longer. This is one of the few metabolic myopathies where enzyme replacement therapy can add to the benefits of standard supportive therapy.

A member's
STORY

Rolling with the punches and dancing to her own trumpet

When Freda Evans was diagnosed with Pompe disease more than 30 years ago she decided then and there that the condition would not rule her life. Anything people said she couldn't do, she has done – including travelling the world alone. *By Melanie Louden.*

Freda Evans was, when diagnosed, the only person with Pompe disease in New Zealand.

When Freda Evans was diagnosed with Pompe disease her reaction was a mix of happiness, sadness and determination.

After suffering bad headaches and weakness in her legs for a couple of years, tests and biopsies were carried out. When the doctor told the then 33-year-old she had Pompe disease, she said: "Okay, so what's the cure?"

"Back then there was nothing, and there's still nothing to this day," says the now 65-year-old, who lives in Auckland.

"I went from being a normal person to being a unique person because I was the only one in New Zealand with Pompe."

Pompe disease is a rare disease that damages muscle and nerve cells throughout the body. It leads to progressive muscle weakness, wheelchair dependence and breathing assistance.

"I felt happy that I had a diagnosis, but sad that I was probably going to die from it, and sad that there was nothing they could do for me, other than keep me comfortable."

There are now 12 people in New Zealand with Pompe disease, including two of Freda's whānau, and the NZ Pompe Association was set up to help people with the condition.

Keeping Freda comfortable involved her being put on overnight ventilation via a Bi Pap machine "from day dot as my lungs were proving to be very unreliable".

However, Freda was not going to let the rare disease take over her life.

"I took the bull by the horns way back then and said 'you're not going to rule my life. I'm going to rule my life. I will dance to my own trumpet'.

"Anything people said I couldn't do, I did. I've travelled around the world alone. I chose to not let it rule my life. But in saying that, it wasn't always easy."

But there is plenty to keep Freda smiling – she has been working as a kōhanga reo administrator since around the time she was diagnosed 32 years ago, and says it is an absolute joy to come to the South Auckland centre each day and see the smiling faces of the children.

She is also a Justice of the Peace and is involved in the marae and kura.

Freda is one of eight siblings, she has two sons and three granddaughters, and says one of her proudest achievements in life is being a mum.

A member's
STORY

Freda says the key to living with Pompe disease is taking life one day at a time.

"If you are a strong person in yourself, you just have to roll with the punches."

"I'm proud of everything. I just get on with life."

Freda says it was hard, at that time, being the only person in the country with Pompe disease.

"It was lonely. You have no idea. No one knew anything about it. No one could help me. I have some idea of what it's like to be an orphan now."

It was 13 years before another Kiwi was diagnosed.

There are now 12 people in New Zealand with Pompe disease, including two of Freda's whānau, and the NZ Pompe Association was set up to help people with the condition keep in touch, to offer support, advice and help.

Freda, and the association are also heavily involved in lobbying the Government to fund Myozyme for late-onset Pompe disease.

A small number of New Zealanders, including Freda, receive the drug as a result of a compassionate fund – but the association wants to see the drug fully funded.

The enzyme replacement therapy is not a cure, but it does give recipients a better quality of life. It can stabilise the disease and prevent deterioration.

The drug is funded for all ages in more than 75 countries and New Zealand is one of just three countries in the OECD that don't fund it for all ages.

However, Pharmac does provide funding for New Zealand infants with Pompe disease – despite the fact no infants in the country have been diagnosed.

Lobbying from the group was fairly quiet this year because of Covid-19 and the elections, but Freda says they will get back into it next year.

"We've just got to keep chipping away at it."

Freda has been "very mindful" of how Covid-19 could affect her health and was in isolation for 12 weeks by herself, except for caregivers who came in to shower her.

She says it was hard, and lonely, but lots of Facetime calls and drive-by visits from whānau helped brighten her days.

Freda says the key to living with Pompe disease is taking life one day at a time.

"If you are a strong person in yourself, you just have to roll with the punches.

"People ask me what I miss. I used to miss wearing high heel shoes. But not anymore. Actually, I don't miss anything. I don't miss playing sports – I get to sit in my chair and watch it on my big TV instead."

Where's my cheeseburger?

BY DR ADRIAN TRENHOLME

Dr Adrian Trenholme reflects on the many special memories he has of the families with children with neuromuscular disorders that he has cared for over the years.

I am a Paediatrician and it has been my great privilege to work with and support families in South Auckland for the past 35 years. What has always impressed me has been the way children and their whanau have accepted the challenges life has given them and then adapted and thrived despite the inevitable periods of anger, grief and frustration.

I have made it a mental routine of mine to think of the families I know when I sit down for my mid-morning cup of coffee on Sundays and reflect on the highs and lows we have shared.

A favourite cartoon of mine is in three boxes –each contains a glass exactly half full of water. The first one is a happy person saying "half full". The second is a scowling person saying "half empty". The third is an average Joe who says "Where's my cheeseburger?".

I like this approach which is positive and challenging and says to me life is about what you can do, not what you can't.

Many of the families I have known have had children with neuromuscular disorders and I have many special memories.

A video of a teenager dancing in a wheelchair and being mesmerizingly good.

A van lowered and customised which was very cool but a nightmare for the parent to drive.

A boy who couldn't talk, struggling to say only two words to me – "fat boy" and then laughing (I think he was talking about the Harley Davidson?).

Ball photos which show me sophisticated beautiful people and photos from trips overseas, happily smiling with Disney characters when I know the sweat and tears needed to get there.

A young lady who, at the age of three was declared "palliative" elsewhere, telling me about her first legal drink of alcohol. Finally, laughter and humour is very much a part of clinic visits and I treasure that.

In the last few years, I have had my own health issues and have had to deal with the health system.

What I have come to realise is that many doctors really don't listen. I value having a GP and a specialist who listen and who I can relate to and see on a regular basis.

They are not always right and they are not afraid to challenge me, but they listen and are trusted access points into the system, rather than the lottery of meeting new people each time.

To me that is the value of regular visits to the Paediatrician – as a connecting point into the system for children with neuromuscular problems.

I like this approach which is positive and challenging and says to me life is about what you can do, not what you can't.

The Covid-19 pandemic has challenged the way we all live and as we all know is of greater risk for those with underlying conditions, especially the elderly.

Interestingly my job has changed dramatically with hospitalisation for respiratory infection in infants dropping dramatically this year.

The consequences of social isolation, caring for elderly and children with underlying conditions at the same time, and job insecurity will impact heavily.

The way we socialise and work

Wash hands, wear masks, reduced contact when you have a respiratory illness, limited travel and wariness of crowded situations.

now has altered to reduce the risk of transmitting respiratory infection to oneself and others.

What we do is just common-sense but has to be done with kind consideration of others, rather than just oneself.

Wash hands, wear masks, reduced contact when you have a respiratory illness, limited travel and wariness of crowded situations.

For families dealing with neuromuscular conditions these are small additional hurdles which I have every confidence they will manage competently and positively.

I wish you all a Merry Christmas – and keep looking for the cheeseburger!

Dr Adrian Trenholme is a Consultant Paediatrician at Kidz First Hospital, Counties Manukau Health.

Giving made simple

Do you like to support worthwhile organisations on a regular basis but don't want the hassle of having to remember to make the payments?

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Let us know once you have put this in place and we will make sure to get you that annual receipt either by mail or email, your choice.

Please visit our website below, enter your details in the contact form and we will work with you to put a payment plan in place.

www.mda.org.nz/Ways-to-Help/Regular-Giving

Thank you. We couldn't do what we do without the support of generous donors like you.

Muscular Dystrophy New Zealand

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

Dr. Tristram Ingham
National Executive Chairperson

Brian Hadley
Accountant and Business Manager

Chris Stichbury
Manager for Research, Development and Monitoring

Melanie Louden
Communications and Marketing Advisor

Shelley Butler
Accounts Assistant

Shanthi Ameratunga
Clinical and Scientific Advisor

Contact Centre

Development Leader: Dene Benham.
Supervisors: Maatuakore Wirihana-Tawake, Samuel Boyd.
Senior Customer Service Representatives: Kelly Williams, Melissa Jamieson, Simone Wareham, Vicky Ferguson.
Customer Service Representatives: Ashia Porteous, Annette Glasglow, Carlos Bennett, Rawiri Clarke, Tamara Sergent.
Administrative Assistant: Janine Gardner.

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

Our branches

Northern Region

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

Central Region

Community Coordinator – East Coast District: *Michelle Smith (left)*
Community Co-ordinator – Wellington Region: *Pip McLean (right)*

Fieldworkers: *Talitha Vial (left)* and *Raygaana Naidoo*
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: mdacanty@xtra.co.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
Michael Schneider
Email: support@mdn.org.nz

Central Branch
Bernadette Ingham
Email: members.central@mda.nz

Canterbury Branch
Mike Nolan
Email: mike.nolan@mda.org.nz

Southern Branch
Tristram Ingham
Email: chairperson@mda.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

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

Vela Tango Independence Chair

The Vela Tango is a unique indoor mobility chair that creates independence for anybody with reduced mobility.

- Walkable and offers stability with a central brake. More functional than a wheelchair or walker for indoor tasks.
- The 20cm height adjustable by an electric switch under the armrest. Allows for a greater reach, functionality and assisted standing.
- The seat can rotate 90° LH and RH. A quick release button will fold back the armrests to accommodate easier transfers to another seat or bed.
- Award winning 2-part ALB backrest with adjustable lumbar support, including tilt and height adjustability.

TOPRO Pegasus Carbon

7 Years Warranty

- Ultra lightweight, rigid carbon fibre frame, with stylish detachable shopping bag and backrest. Additional helpful accessories available.
- The rollator can be collapsed with one hand and secured with the lock clamp for transport or storage.
- The unique edge guard feature prevents the walker from catching on furniture legs and doorframes. The tilt assistance function gives the foot a wide area to step on and tilt the walker up to navigate kerbs.

TA Electric Toilet Lift

- A high quality vertical lifter designed to assist people with weak leg muscles to stand and sit unassisted from a normal toilet.
- 40cm of variable lifting interval provides increased independence for the user.

TOPRO Taurus Electric Walker

- The TOPRO Taurus has electric height adjustment by a simple push of a button and can assist some users from sit to stand.
- Adjustable forearm supports, and a wide range of accessories make this the ideal walking aid for users with restricted mobility.
- Suitable for users 140cm – 210cm.
- 6 Optional accessories available.

Obi Independent Dining Robot

- With the touch of a switch, Obi allows users to select between four compartments of food and to command when the food is delivered to their mouth.
- Obi increases independence, social interaction, and effective food capturing like never before!
- Obi is perfect for eating out! Obi is portable, 3Kg and provides up to 3 hours of eating time per full charge.
- #1 Occupational Therapist Recommended Robotic Feeding Device in the U.S.

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