InTouch

KIA NOHO TATA Inclusive | Inspiring | Informative. Winter 18 Issue 99

Achieving life goals

Our first Dukies go for bronze

Working through grief

A very personal process

Circulation boosters

Keeping your veins healthy

Managing pain

Understanding the options

Muscular Dystrophy New Zealand

InTouch

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Korero with Ken

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

My special thanks to those people who have stepped up to become members of our branch committees and national council. I would also like to express my gratitude to those that have stood down, their contribution to MDANZ is really appreciated.

For those who missed the AGM, I would like to highlight some of the great achievements over the last year. Our membership has grown by 118, more members are taking advantage of the pneumonia vaccine, there were 48 successful applicants to the member's discretionary fund, 120 members and families attended educational events about their specific neuromuscular condition, and there were a staggering 16,800 copies of *In Touch* magazine circulated. All this was achieved with the group achieving a small net surplus of $2,668. Congratulations to all the team for this great outcome.

The AGMs around the country were an opportunity for both doing business and catching up with friends.

I also want to acknowledge Val Abrahams who recently passed away. Val was a life member of MDANZ and was instrumental in helping to form the organisation in the 1960's. She served 33 years on National Council. On behalf of MDANZ, I would like to express my condolences to Val's family.

As the year marches on, winter time is again upon us. I find it a challenge dealing with the cold and the limited opportunities to get out and about. Yes, I have had my flu vaccination and had my pneumonia vaccine last year. And I have a warm, dry home, companions to warm the heart and a good supply of Shiraz for long nights in. On reflection, what have I got to moan about? One hundred years ago this year my grandfather was stationed on the Western Front as one of the more than 100,000 men and women serving in World War One. The challenges they faced, make mine pale by comparison. E mihi nui ki nga tupuna o tenei motu ataahua, moe mai – greetings to the ancestors of this beautiful nation, sleep well.

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

Ken

Ken Green
MDANZ Chairperson

In touch with Ronelle

Ngā mihi nui ki a koutou, warm greetings to you all.

The past few months can be summarised under our key organisational value Connected – Tūhonotanga. This reflects how we value the role of whānau and communities and invest time and resources to foster both collective awareness, and strong relationships.

It has been an amazing time of connection and I have been busy representing MDANZ and our members at a wide variety of events, meetings, forums and conferences. Each of these hui (gatherings) have a different focus and my role varies according to whether I am hosting an event, presenting information at a conference, or participating as an audience member.

I travelled to Wellington to represent MDANZ at the NZ Disability Sector Network conference, DSS Consumer Consortium, Health Quality and Safety Commission conference, Ministry of Health NGO symposium and Carers Alliance meeting.

During one of these trips I attended parliament to present a submission to the Health Select Committee on the Misuse of Drugs Act (Medicinal Cannabis) Amendment Bill, to raise the profile of our community and advocate for improved access to a wider range of safe, regulated products for managing pain. Pain is an issue for many of us – 60% of adult participants in the MD-Prev study reported pain as a key symptom of their condition. For this reason, I'm also encouraging the members of the long-term conditions planning team at the Ministry of Health to include pain as a quality of life indicator in their framework. You can read more about pain management on page 31 of this issue, with Miriam Hanna offering information and ideas.

Collaboration across our rare disorders community continues, with examples such as support for Rare Disease Day, the NZ Pompe Network Conference and the recent campaigning for medicines access for Spinal Muscular Atrophy (SMA). The SMA Member's Reference group is proving to be a fantastic initiative, as paid staff at MDANZ work together on shared priorities with SMA community members in a partnership model.

It was great to see members and supporters attend AGMs around the country and I'm grateful to our community members who have stepped forward to participate in governance for the coming year. It is a vital role for our organisation. A special thanks to Disability Rights Commissioner Paula Tesoriero, who spoke at the Wellington Branch AGM. We are enjoying getting to know you in your role.

Each point of connection we have is an opportunity to create meaning and impact.

Hope you enjoy this issue of *In Touch*.

Hei konā rā, Bye for now.

Ronelle Baker
Chief Executive

Outside Parliament with members of the Carers Alliance.

A cup of tea and a catch up with … Rachel Woodworth

Each issue we introduce a MDANZ team member:

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

I joined the team in April and am a fieldworker for Northern Branch covering central Auckland and Northland. Before this job, I worked as a community transport coordinator for Auckland Transport; and as an injury prevention consultant for ACC for eight years. I have also worked as a specialist Orthopaedic/Trauma registered nurse working in the UK, Australia and here in New Zealand in Whangarei.

What qualifies as a great day at work for you?

Variety, but mainly meeting people face-to-face, making meaningful relationships and making a positive difference in their lives, however small that may be.

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

I would like to see our members taking advantage of every positive experience and opportunity that comes their way.

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

Cheese scones with black cherry jam, and a cup of steaming hot green tea in a china cup and saucer.

What are you passionate about?

I am passionate about life and try to enhance the lives of people that I know or who are around me in a positive way; we only get one shot at it so I do my best to try or experience something new or different every day.

I am passionate about being the best mother I can be to my 19-year-old son Billy.

I am passionate about music and dance; music is such a dictator of mood and dance is under-rated.

Save the date

June 20: World FSHD Day.

September: Freedom Campaign. Talk to your branch to find out how you can get involved.

September 15 – 16: Friedreich's Ataxia Forum day, Brentwood Hotel, Wellington. Check the FARA NZ website for more details.

Support us!

Any donation, big or small makes a difference. Donations of $5 or more are tax deductible.

Call: 0900 426 93 to make an automatic $15 donation.

Online: Donate any amount securely online. **www.mda.org.nz**

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

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

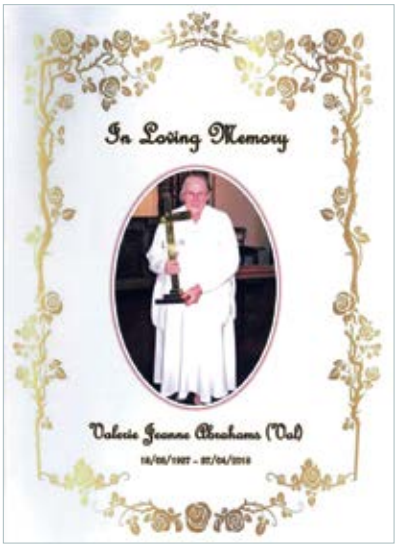
Thank you. We greatly appreciate your support.

Remembering Val Abrahams

'Ka hinga te tōtara o te wao nui o Tāne' (a mighty tōtara in Tāne's forest has fallen) is a Maori proverb quoted when someone of importance passes away.

We offer this proverb as a tribute to MDANZ life member Valerie Jean Abrahams, known by us as Val. Val played a vital role in helping to establish our organisation in the 1960's, providing 33 years of service at branch and executive levels of organisational leadership, while fundraising and fostering social interaction for our community. Val was recognised in 2002 and made a Member of the New Zealand Order of Merit for her contribution.

Val was the main carer for her husband and daughter who both had Myotonic Dystrophy. At her funeral service John Adams, another MDANZ life member, spoke about how he and his wife met Val and her husband Eric at an AGM in 1971. This was a seminal moment for the Adams to become involved with the organisation, as their two sons had just been diagnosed with Muscular Dystrophy.



John's historical tribute, and Val's passing is a timely reminder as we approach our 60th Jubilee, of the many people whose hard work set the foundation for this amazing member-led organisation that we now have stewardship over. We express our gratitude and our sympathies to the Abrahams family at this time.

Seeking back issues

We like to keep a library of back issues of *In Touch* and are very low on copies of the Winter 2016 issue. If you've got one spare, we'd love to add it to our archives. Please email info@mda.org.nz and we'll arrange postage.

From the branches

Northern

Thanks to Sue Field and Andy Dowdle who did Tour Aotearoa, a 3,000km bike ride from Cape Reinga to Bluff, and raised more than $2,000 towards the next MDN Family Camp to be held from 23rd-25th November at Ngaruawahia.

Wellington

Members often mention they would like more opportunities to get together, so Wellington-based fieldworker Dympna, will be hosting a coffee morning soon. If you're interested, let her know on 0800 886 626 or email dympna@mda.org.nz. Any ideas on where to hold it would be most welcome.

Canterbury

The annual youth camp is one of Canterbury Branch's most highly anticipated events and this year they are inviting all members aged eight to 18 to come and join in the fun at Mt Hutt Retreat, Methven from October 1st-4th. Catch up with old friends and make new ones. Let your fieldworker know if you are interested, or contact Bonny Stephens.

Southern

A LEGO group is about to start soon. If your children are interested, or you would like to help out, please contact fieldworker Jo at joanne@mda.org.nz.

Great day out for SMA families

Families travelled from as far away as Christchurch for our SMA Family Day held in Hamilton.

The April event was a fantastic opportunity for our members with Spinal Muscular Atrophy (SMA) to mingle and get to know each other, provide support and share information. The event was sponsored by Biogen, and wouldn't have been possible without a grant from them.

The children had a fantastic time, as The Link Community Centre was accessible with an outdoor playground for children to be entertained. Zebra soft toys, sponsored by Valerion, were given to each family, along with the book Zac's Play Day, sponsored by Biogen. Zebras symbolise rare disease as every zebra's stripes are unique with no two zebras the same.

The group heard from a variety of speakers. Gina O'Grady, paediatric neurologist from Starship, gave an overview of SMA and shared the exciting news of the Spinraza Expanded Access Programme soon to be offered at Starship Children's Hospital for eligible children with Type 1 SMA. Miriam Rodrigues spoke about the NZ Neuromuscular Disease Registry and the Global SMA post-marketing surveillance registry, set up to provide further information following the introduction of new treatments such as Spinraza.

The children loved the toy zebras and new book.

Families also heard from Carol Armstrong, of Sports Waikato, who gave interactive demonstrations of inclusive sports such as Boccia.

Niki Russell, dietitian from Nutrition Care, discussed common dietary and nutrition issues in chronic conditions such as SMA, and answered several questions from the group. Cynthia Ward from True Colours discussed coping, and the importance of self-care and compassion, and the role of counselling and support organisations. Lastly, the SMA member's reference group were introduced and MDANZ chief executive Ronelle Baker gave an overview of the New Zealand political landscape, and our plans for working with the SMA community on advocacy priorities. N

Self-care Sunday

More than 60 people got together to drink tea and enjoy yummy petite treats at our sold-out High Tea, held at historic Ferndale House in Auckland's Mt Albert at the end of March.

As well as catching up with their friends, guests were treated to the wisdom of psychologist Anna Friis, who spoke about the importance of self-compassion and how we need to be just as nice to ourselves as we are to the other people in our lives.

It was great to have patron Judy Bailey and Carmel Sepuloni, the Minister for Social Development and Disability Issues, join us for the afternoon.

We are very grateful for the support received from members of our community who donated food, raffle items, and helped out behind the scenes. N

Above: CE Ronelle Baker with Minister Carmel Sepuloni. Above right: Thanks to Maree Biland (centre) and other volunteers who made the day a great success

Understanding SCA and FA

Using nerve ultrasound as an investigative tool.

The aim of Dr Luciana Pelosi's research, funded by Neuromuscular Research New Zealand, is to gain a better understanding of the underlying pathological workings of the sensory and motor nerves in spinocerebellar ataxia (SCA) and Friedreich's Ataxia (FA), by using a new tool, nerve ultrasound.

In recent years, nerve ultrasound has emerged as a useful tool for the investigation of peripheral neuropathy, showing abnormal nerve enlargement in demyelinating neuropathies and, to a lesser extent also in axonal neuropathy.

By using this technique, Luciana and her team have shown that patients with cerebellar ataxia neuropathy vestibular areflexia syndrome (CANVAS), who have sensory impairment from sensory neuronopathy, have pathologically small nerves on ultrasound. Small nerve size is consistent with nerve thinning from axonal loss secondary to ganglion cell death. This ultrasound finding in CANVAS was unique and significantly different from that in a group of age and gender matched patients with 'axonal' neuropathy, whose nerves were enlarged.

Luciana's preliminary nerve ultrasound study in people with Friedreichs's ataxia showed that peripheral nerves are enlarged in this condition, especially at proximal upper limb level. This was somewhat unexpected, as the sensory impairment in this condition has been generally attributed to dorsal root neuronopathy. However, the ultrasound finding is consistent with recent studies suggesting an independent abnormality in the peripheral nerves.

Her pilot study of nerve ultrasound in 12 individuals with SCA has shown abnormalities of different types in different SCA subtypes, with small nerves consistent with neuronopathy for SCA1, SCA2 and SCA3 and, large nerves consistent with 'axonal' neuropathy, in SCA6 participants. Interestingly, ultrasound appeared to be more sensitive in identifying abnormality than nerve conduction studies.

Luciana will contextualise the ultrasound recordings in people with SAC and FA by comparing them with clinical disability scales, clinical sensory testing and nerve conduction studies. This research may help us better understand these conditions as well as providing a useful progression marker for clinical trials in FA and SCA.

This line of research was initiated by Luciana Pelosi with colleagues in New Zealand, including Eoin Mulroy, Richard Roxburgh, Miriam Rodrigues and Dean Kilfoyle, and, as no other overseas research groups have their level of experience with nerve ultrasound, Luciana is uniquely positioned to pursue this definitive study. This will also provide a major collaborative opportunity between New Zealand neurology and international ataxia research groups.

Dr Luciana Pelosi.

Getting together at National Office

A night for business, stories, and special acknowledgements.

The National AGM was held 20th April at National Office in Penrose Auckland and made available via web-conferencing for members and staff to join remotely.

Members and supporters were in attendance, including special guest Andy Sinclair General Manager of Hyundai NZ, former National Council Chairperson Helen Melrose and outgoing Rangatahi Representative Olivia Shivas.

Ken Green presented his Chairperson's Report, sharing a treasured family heirloom, a bugle given to his Grandfather during World War 1, from his fellow bugler who served in the New Zealand Rifle Brigade. The Bible refers to blowing trumpets to summon the people and to break camp, and Ken used this as a metaphor of calling our members not only to the AGM but to the organisation. He also made reference to the Maori contingent who inscribed on the bugle 'Kia kaha, kia toa', be strong be brave. He added a third message 'kia manawanui', reminding the organisation and its leaders to be strong, be brave, be steadfast.

Ken reiterated that MDANZ is only here for its members and that they are the highest priority. He acknowledged the significant increase in member engagement this year, through the fieldworkers, branches and events held for members, many of which he has attended.

Brian Hadley presented the financial report, and the final group result saw a $2668 surplus for the year. This is a great result and significant turnaround from the previous year.

Ronelle Baker presented the Chief Executive's report, expressing pride in what MDANZ have achieved, with hard work and a goal oriented approach by the team. She acknowledged the mana enhancing stories of our members that reflect who we are and what we stand for. She signaled that we need to continue to live our core values and develop our organisational infrastructure with robustness to provide quality assurance to our stakeholders. We also need to continue to provide opportunities to bring people together – this is where the magic happens.

The outcome of the National Council elections was shared. Three nominations were received for the three vacant positions available, therefore the nominees were elected unopposed and no vote was needed.

Elected councillors are:

Scott Laurenson – Councillor at Large

Brent Walker – Councillor at Large

Scott Boyle – Rangatahi Representative

Also noted that Andrew Willetts replaced Robbie Verhoef as Southern Branch Representative and Andrew Munro will replace Warren Hall as Canterbury Branch Representative.

Formal business was followed by engaging presentations by Shelby Taylor and Miriam Hanna – both recipients of a Post Graduate scholarship from our research trust, Tonya Baker speaking about the Duke of Edinburgh's International Hillary Award programme and Russell Just speaking about the fundraising partnership between Community Power and MDANZ.

Attendees at the National Office AGM were inspired by a piece of history – Chair Ken Green bought along the bugle his grandfather was given during World War 1.

AGM news

Northern

The Wellington Branch AGM was held at the National Office in Penrose Auckland, on Sunday 15th April. Fieldworker Darian Smith presented about his role and the strong alignment with MDANZs organisational Vision of Freedom beyond limits. Tonya Baker also talked to the group about the Duke of Edinburgh's International Hillary Award.

Trevor Jenkin was re-elected as Branch Chairperson. Michael Schneider continues as branch representative on National Council. Andrea Clive was re-elected as Treasurer and Joy Jenkin continues in the role of Secretary. New committee members were welcomed and a farewell noted to Rebecca Poad who has moved out of the area and will now serve on the Canterbury Branch Committee.

Wellington

The Wellington Branch AGM was held at the Petone Community Centre, Saturday April 7th and members enjoyed hearing from guest speakers Dane Dougan (CE Autism NZ) and Disability Rights Commissioner Paula Tesoriero.

Annelize Steyn was re-elected as Branch Chairperson and continues as branch representative on National Council. Urmit Patel was re-elected as Treasurer and Bernadette Ingham was appointed to the role of Secretary following the AGM. New committee members were welcomed and those standing down were thanked with a small gift.

Disability Rights Commissioner Paula Tesoriero with CE Ronelle Baker and members of the Wellington Branch Committee (top) and Fieldworker Dympna Mulroy (above).

Canterbury

The Canterbury Branch AGM was held at the Papanui Library in Christchurch on Saturday 24th March and attended by members from near and far.

A minute of silence was held at the start of the meeting to acknowledge longstanding committee member and supporter, Earle Mason who passed away last June.

Andrew Munro was re-elected as Branch Chairperson and appointed as branch representative on National Council. Warren Hall has been re-elected as Vice Chairperson and Rebecca McClean as Treasurer. Acknowledgements were given to committee members who stood down this year. A number of committee members were re-elected and some new members came on board.

Guest speaker Tonya Baker talked to the group about the Duke of Edinburgh's International Hillary Award and committee member Bonny Stephens gave a presentation about last year's very successful Youth Camp, which will be repeated again this year from 1–4 October.

Southern

The Southern Branch AGM was held at the new office premises, Cargill Enterprises, in South Dunedin on Saturday 10th March. Robbie Verhoef stood down as Chairperson, and was replaced in the role by Andrew Willetts who will also take up the role of branch representative on National Council. Raewyn Hodgson was re-elected as Secretary and Rebecca Croxen as Treasurer. Committee members were re-elected for another term. CE Ronelle Baker joined the meeting by Skype to present information on organisational priorities and member services.

Ella Mills. Photo credit: The Artset.

Our first five

Catching up with our Dukies

Our first cohort is well underway to achieving bronze in The Duke of Edinburgh's Hillary Award

Ella Mills – Waiuku, Auckland

So far, I have completed: Badminton (Physical), hairdressing (Skill)

The part I have enjoyed the most has been: Volunteering at the RDA (Riding for the disabled). The time and effort they put into helping the kids is incredible.

The most challenging part of the programme for me has been: Playing badminton because I get tired very easily. But it helped me develop new strategies to adjust the way I handle my tiredness.

Something I have learnt about myself is: I am self-motivated, once I set a goal I will finish it no matter what it takes.

I would recommend this programme to anyone who: Wants to learn new skills.

Jack Lovett-Hurst – Invercargill

So far, I have completed: Radio FM Assist (Service), cooking (Skill), cycling New York Marathon (Physical)

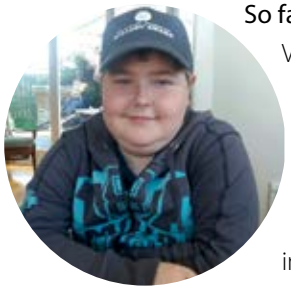
The part I have enjoyed the most has been: Achieving different goals.

The most challenging part of the programme for me has been: Setting all the goals at once and completing these whilst training for the New York Marathon.

Something I have learnt about myself is: How well I coped with travelling across America, and got on and completed the goal I had set for myself.

I would recommend this programme to anyone who: Would like to try something different and achieve goals that they never done before.

Dylan Schneider – Auckland



So far, I have completed: Vex IQ Robotics (Skill), Air rifle shooting (Physical).

The part I have enjoyed the most has been: Air rifle shooting, it was fun, interesting and I met lots of nice people.

The most challenging part of the programme for me has been: Doing the robotics as a lot of work was put into it and it got very tiring at times. But I still carried on and completed it.

Something I have learnt about myself is: I didn't know if I could do it. I didn't know I was persistent, but I discovered I was very persistent with my goals.

I would recommend this programme to anyone who: Wants to learn new things and isn't doing anything like this at present.

Grace Chapman – New Plymouth

So far, I have completed: Puzzle making (Service), and am part-way through my cake decorating (Skill).

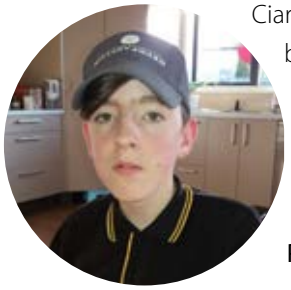
The part I have enjoyed the most has been: Eating the cakes I am decorating!

The most challenging part of the programme for me has been: Trying to do all the different goals and staying on task.

Something I have learnt about myself is: I am not very patient at times, especially when I am trying to decorate my cakes.

I would recommend this programme to anyone who: Wants to give it a go.

Ciaran Calder – Nelson



Ciaran is waiting for surgery before pursuing his goals.

Service: Volunteering with dogs

Skill: Rifle shooting

Physical: Table tennis

We're recruiting!

Award Leader Marty Price is looking for another five young people, aged between 14 and 24, to begin the bronze level programme this year. To find out more and receive an application form, email marty@mda.org.nz.

What is involved in completing the programme?

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

They choose a:

- **Service** – For our young people who may rely on the assistance of others at times, the ability to contribute and give service is highly empowering, and great for their sense of self-worth.
- **Physical Recreation** – This can be as much or as little as an individual is capable of. It's about stretching themselves to achieve.
- **Skills activity** – This could be an existing skill, or something new – gardening, fishing, baking etc.
- **Go on an Adventurous Journey** – Plan and complete a trip away from home.
- **To achieve a Gold Award.** Take part in a Residential Project.

Understanding grief

A personal process that's different for everyone

The experience of grief often requires us to embark on a personal journey, learning new things about ourselves and different ways to view the world.

A diagnosis of a life-limiting condition often brings a period of grief, and this can recur throughout life as the condition progresses. There is a sense of travelling down a dividing line between the past and the future. Looking backward reminds us of things that have changed or been lost. Looking forward, it's hard to visualise a different life and to stay positive. There's no going back to the past and the future is uncertain. It is therefore no surprise that grief can be overwhelming, challenging and life changing.

What is grief?

Grief is the human response to change and loss in our lives. It is a natural and normal response, which can have a physical impact on our bodies as well as affecting our thoughts, emotions and mood. Grief challenges the way we think about ourselves and the world, and therefore can have an impact on our relationships with others. Our grieving process can also be influenced by our cultural and spiritual values and beliefs.

Each of us expresses grief in a different way. Some people openly express their grief and pain, while others may withdraw. They may not cry as they try not to upset others around them. It is common for someone close to a person who is struggling with change to also find themselves in their own grief process. Relationships may become a focus, as people are reminded of how their relationship and lives used to be.

It is important to remember that grief is a normal and natural response and that there is no right or wrong way to grieve.

Emotions you may feel…

- Anxiousness
- Disbelief or confusion
- Guilt
- Anger, frustration or disillusionment
- Loneliness
- Isolation
- Consumed by sadness

Physical reactions you may experience…

- Sleep disturbances - either too much or too little
- Changes to appetite or eating habits
- Digestive problems
- Tightness in chest, throat and breathlessness
- Decrease in energy levels, e.g. feeling tired or lethargic
- Anxiety and panic attacks

While grieving is a personal process that has no time limit, nor one "right" way, there are some universal principles that can be applied. Professor of Psychology, J William Worden, has identified four key steps or tasks that can be worked through for a wide range of losses we grieve for in life;

1. To accept the reality of the loss.
2. To process the pain of grief.
3. To adjust to a changed world after the loss.
4. To find an enduring connection with what has been lost whilst embarking on a new life.

These steps do not necessarily need to be approached in a linear way and it is likely that you will revisit and reprocess each task several times as time goes by. It is important however to give each task time, and engage in the process in ways that feel right to you.

Moving through grief

Understanding the grief process is an important step towards adjusting to change in your life and can help to make the grief journey less bewildering. It can be helpful to begin by identifying and honestly acknowledging each emotion that you feel.

While grieving is a personal process that has no time limit, nor one "right" way, there are some universal principles that can be applied.

Don't ignore or push away uncomfortable feelings, allow them to 'be,' and then direct your attention towards considering how best to manage them when they arise. Think about what strategies you might create for yourself for when your feelings become overwhelming. You could discuss these strategies with your family and friends, counsellor, GP, or your MDA fieldworker.

Identifying your feelings may help give you the energy and awareness to take on another step. Take this time to think about focusing on the areas of your life that you can influence rather than those you can't. By not focusing on the 'if only,' you will be able to direct your efforts and emotional energy towards the choices, actions and decisions that will help you to live your life in a way that is meaningful to you.

Creating space in your life and finding extra support to work through grief may be helpful. This may include:

- Time out from your responsibilities to focus on your grief or to refresh yourself
- Finding spaces or places where you can safely experience different emotions (such as sadness, anger, fear, guilt and humour)
- Talking with a friend, family member or fieldworker who you can talk openly with to help make sense of what has happened and help you reflect upon your reactions without feeling judged
- Building a support network of people you can call upon to help in a range of contexts including the practical aspects of daily life – with the mindset that it is okay to ask for and accept offers of help from others

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RESPIRATORY TECHNOLOGIES

- Writing in a journal to bring your thoughts and feelings to the surface, help with reflection and gaining new perspectives
- Meal planning and buying in nutritious food to nourish your body while it processes difficult emotions
- Doing any form of exercise to help with physical symptoms and manage your mood
- Practising meditation or mindfulness to help manage strong emotions and offer clarity of focus on the present
- Professional counselling or psychotherapy to work through feelings such as sadness, fear or anxiety

Be kind to yourself and don't be afraid to ask for the support that would be most helpful to you. If finding time for yourself is a challenge, ask someone to look after the children for an afternoon. If a messy house is compounding your grief, ask a relative to give you a hand tidying up. Your fieldworker could help you identify your support network and help you to work through some practical options or strategies, to help you acknowledge and move through grief. There are also professional supports and online resources you can access.

Extra support

A free confidential counselling service is available for MDANZ Members with a neuromuscular condition and those close to them. It's quick and easy to access this counselling programme. Phone 0800 SELF HELP (0800 735 343) and give your name and MDANZ membership number. You will be referred to a professional, accredited and experienced counsellor in your area.

For more information about grief visit the Good Grief website at www.goodgrief.org.au

Depression.org.nz has some practical tips and an online journal to help with staying positive and managing mood.

Find out about mindful self compassion at www.centerformsc.org or look into meditation http://how-to-meditate.org/

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Taming the dragon

Sharing my story

After going through a rare Myasthenia Gravis crisis, Mereti Taipana-Howe wrote a book to share her story of overcoming seemingly insurmountable odds.

Five months in hospital took a lot of things away from Mereti Taipana-Howe, but she has used it as a way to give back, writing a story that is very personal, but also a gift to those who share her condition, and their families.

Mereti says her book *Taming the MG Dragon: Journey through a Myasthenic Crisis* allows the reader to do three things.

"One to learn more about the disease Myasthenia Gravis (MG), two; to get a rare insight into a severe life-threatening crisis within the disease, and three; learn how a person can recuperate from such a terrible experience," she says. "It is not a medical text, but it could prove useful for medical professionals, social workers, counsellors as well as those with the condition, and their whānau."

Her own family members encouraged Mereti to start writing about her experience during her five-month stay in Palmerston North Hospital in 2016.

"At first I did not want to talk about it, I just couldn't go over what had happened. But as I began to improve during the second half of my third month in the Intensive Care Unit (ICU), I decided I would put something together. Then it was a case of deciding how I would do it, and why.

One reason was to process what had happened in my own mind. Another was to help others going through their own difficult situations, encouraging them to see the light at the end of the tunnel. I approached it rationally, rather than emotionally, because I knew the readership would be varied and diverse. I wanted them to come away inspired, or at least with some learning."

Despite never having done any formal writing before, Mereti discovered a new passion for storytelling, although it wasn't always an easy process.

"It took a year and a half to complete," she says." It was a difficult process because I was in ICU for three

One reason was to process what had happened in my own mind. Another was to help others going through their own difficult situations, encouraging them to see the light at the end of the tunnel.

months of that time, unable to talk, swallow, or write because my hands did not work. After about two and a half months I was able to start taking down notes. Some of the information came from doctors, nurses and my whānau. I didn't have any memories of some of the things that happened, such as having a cardiac arrest, being resuscitated, being intubated, and other difficult times in the ICU."

The title of the book is a direct reflection on what the experience was like for Mereti.

"The experience of being in the grip of an MG crisis at its most severe, was like being hunted by an all-encompassing fire-breathing dragon. It was also about how I recovered from this crisis, and how, through medical weaponry and whānau support, the 'dragon' was tamed."

There has been fantastic feedback from readers all over the world, and Mereti is very grateful to those who encouraged her along the way.

"I'm proud I am able to talk about such a traumatic experience, which in itself is not easy to do. My story is that one can recover from a severe illness, and it's important not to give up. I'm proud that I continued right through to the completion of the book. It was very hard, and I did not know how to go about publishing it. I got help from unknown quarters and managed to put it together. I feel it was a great achievement, and I am humbled by all those who were with me during my illness, and those who helped me get the book together.

Mereti also has a message for those who may be struggling with their own illness.

"If you end up in hospital, try not to be frightened. Just hold on with all your might and you will get through."

Taming the MG Dragon: Journey through a Myasthenic Crisis is available online through Fishpond and Amazon, at Unity Books in Auckland and Wellington, and at Bruce McKenzie Booksellers in Palmerston North. You can also order it from your local bookshop, or find it at your library.

Another door opens

Member Bryan Beames shares how he made sure his late wife's forgotten story was published.

Although I was diagnosed with Spinocerebellar Ataxia (SCA) some seven or eight years ago, and learned to live with it and its limitations, my darling late wife's story is much more interesting. I also learned to live in her shadow, so to speak.

We were both born in Oxford, England and married in 1958. Margaret was a teacher and I was a ground engineer in the RAF. When my contract to serve for 22 years came to a conclusion we decided to emigrate to New Zealand.

Margaret tried to get into teaching (she was brilliant with young children) but the government decided she would need to spend a year in Christchurch to be re-trained. We both considered this unnecessary and unacceptable. She recalled, as a child, writing stories for her own amusement and thought she would try writing a story for publication. Her first novel, *The Greenstone Summer*, was accepted immediately by publishers both in the UK and in New Zealand. Forty-one books later and after her untimely death in 2016, I was shredding reams of A4 paper, (she was a prolific writer) when I came across a completed manuscript that had apparently never been offered for publication. For a long time I was undecided what to do with it and finally sent it to a childrens' book reviewer Barbara Murison to see what she thought. The reaction from her was swift – "I couldn't put it down" and, "It must be published", so the die was cast. *The Owen Girl* has now been published.

So I said, when addressing the Probus Club of Feilding that 'When one door closes, another one opens – never give up'. *Bryan Beames*

A guide to healthy circulation

Find out more about the signs, symptoms and risks

Movement, hydration and elevation are all keys to preventing a serious blood circulation problem, known as venous insufficiency.

What is venous insufficiency?

Arteries carry blood from the heart to the rest of the body, and veins carry blood from the limbs such as the legs back to the heart. Valves in the veins stop the blood from flowing backward. When these valves are not functioning as they should, and blood starts to pool and collect in the veins, this is known as venous insufficiency.

Several factors can cause venous insufficiency, though it's most commonly caused by blood clots (deep vein thrombosis) and varicose veins. When forward flow through the veins is obstructed — such as in the case of a blood clot — blood builds up below the clot, which can lead to venous insufficiency. In varicose veins, the valves are often missing or impaired and blood leaks back through the damaged valves. In some cases, weakness in the leg muscles such as with many types of muscular dystrophy, can also contribute to venous insufficiency, as the muscles aren't able to effectively squeeze the blood up.

Venous insufficiency is more common in women than in men. Risk factors include an age of over 50, previous pregnancies, smoking, cancer, inactivity for prolonged periods of time (a common factor for our members with mobility difficulties), and family history.

What are the symptoms?

Symptoms of venous insufficiency include: swelling of the legs or ankles (oedema), pain that gets worse upon standing and gets better when legs are raised, leg cramps, aching, throbbing, or a feeling of heaviness or tightness in the legs, itchy legs, weak legs, thickening of the skin on the legs or ankles which sometimes has a leather-like appearance, skin that is changing color such as to a reddish-brown tone especially around the ankles, leg ulcers and varicose veins.

How is it diagnosed?

A physical examination and medical history are generally the first steps to diagnosis. Further testing is likely to include a venogram or a duplex ultrasound. During a venogram, an intravenous (IV) contrast dye is injected into the veins to cause the blood vessels to appear opaque on an X-ray image making them more visible. A duplex ultrasound may be used to test the speed and direction of blood flow in the veins. This is non-invasive and a hand-held device (transducer) is used that uses sound waves that bounce back to a computer and produce the images of blood flow.

How is venous insufficiency treated?

Treatment will depend on many factors, including the cause, health status, history, symptoms and severity, age and tolerability of medication and procedures. The most common treatment for venous insufficiency is prescription compression stockings. These special elastic stockings that apply pressure at the ankle and lower leg. They help improve blood flow and can reduce leg swelling. Compression stockings come in a range of strengths and lengths.

Treatment for venous insufficiency can include several different strategies:

1. **Improving blood flow:** Keeping legs elevated whenever possible, wearing compression stockings, keeping legs uncrossed when seated, and regular exercise will all help improve blood flow. Wheelchair users may benefit from an Occupational Therapist consultation to discuss postural management, seating and pressure relief that may support better blood flow.

When these valves are not functioning as they should, and blood starts to pool and collect in the veins, this is known as venous insufficiency.

2. **Medications:** These include diuretics, anticoagulants and vasodilators. Diuretics draw extra fluid from your body that is then excreted through your kidneys, anticoagulants thin the blood and vasodilators such as pentoxifylline (Trental) helps improve blood flow by widening vessels.
3. **Surgery:** This can include surgical repair of veins or valves, removing (stripping) the damaged vein, endovenous thermal ablation which is a newer method using high-frequency radio waves or a laser to heat and close the problem vein, ligation or minimally invasive endoscopic surgery to tie off varicose veins, vein transplants and vein bypasses where a healthy vein is transplanted from somewhere else in your body in very severe cases.
4. **Ambulatory phlebotomy:** This outpatient procedure where varicose veins are pulled through small surgical incisions under local anesthetic.
5. **Sclerotherapy:** This procedure requires a chemical to be injected into small to medium sized damaged veins so that they're no longer able to carry blood. Blood will return to the heart through other veins, and the damaged vein will eventually be absorbed by the body.
6. **Catheter procedures:** In severe cases involving larger veins, a catheter is inserted into the vein, with heat applied to the end of it, and then removed. The heat will cause the vein to close and seal as the catheter is taken out.

How to prevent venous insufficiency

If there is a family history of venous insufficiency, or increased risk due to loss of mobility from muscle

Walking (whether aided or unaided), and swimming, are effective ways to improve leg strength and boost blood flow.

weakness, steps can be taken to lessen chances of developing the condition:

- Keep moving as much as you can. Try not to sit or stand in one position for long stretches of time. When having to sit for a long period of time, it is important to continue to stretch the legs, wiggle toes and feet and rotate ankles. If having to stand for prolonged periods, it is important to take breaks and elevate the feet.
- For wheelchair users, any movement or leg elevation will help. A support person can help with manual stretches and flexion of your feet, ankles and lower legs. Other devices such as an inflatable foot rest or circulation booster may be used for daily relief.
- Avoid smoking as it constricts blood flow. It is highly recommended that smokers consider accessing support to quit and reduce their risk of venous insufficiency.
- Regular exercise is recommended to help pump blood around the body. Walking (whether aided or unaided), and swimming, are effective ways to improve leg strength and boost blood flow.
- Maintaining a healthy body weight will also decrease your risk.
- Although it may seem counterintuitive when you have swollen legs caused by fluid retention, we recommend staying hydrated with water to aid blood flow and to help your kidneys get rid of toxins in the body.

There are several pieces of equipment that are easy to find and use around the home or when travelling that could help boost circulation. For example, an exercise rubber band that you can buy or request from a physiotherapist may be useful, if you have upper body strength. You can use your arms to hold the two ends of this long rubber band and place one foot at a time through the loop, bending and stretching your leg out in front of you to move fluids and keep blood flowing. You can do this from a seated position in a chair or lying down in bed.

An Aircycle is an inflatable cushion that you place underneath your feet and move the sole of your feet to alternate pressure from the soles to your heels in wave-like motion. This is great for travelling when you can't stretch your legs out in front of you.

If you're at home, a circulation booster that is plugged into the wall is also easy and simple to use. By placing your feet on it, the machine sends out electric stimulation signals that aid lower leg muscles to contract and help blood flow up to avoid blood and fluid pooling. Not all people will be able to tolerate this, so checking with your GP might be worthwhile. We recommend monitoring any pain or side effects carefully.

A simple free standing foot cycle also helps boost circulation and is easy to use in a seated position at home. Note these can also be used for your arms if placed on a table surface at the right height.

Compression stockings are also important to have if you are travelling or expecting to be seated for prolonged periods of time. It is important to find the right size, length, width and tension.

You may want to discuss these products with your doctor or a physiotherapist the next time you see them. Most products can be purchased online (e.g. from ILS https://www.ilsnz.org/Products/Wellbeing/Exercise), and Miriam Hanna at MDA's National Office will be happy to help if you have trouble finding anything.

Golodirsen for DMD

Increasing dystrophin production.

A potential next-generation therapy, golodirsen (SRP-4053) produced by Sarepta Therapeutics, has been shown to increase dystrophin production in Duchenne muscular dystrophy (DMD), specifically for individuals with a genetic mutation that is amenable to exon 53 skipping.

Golodirsen is the second exon skipping therapy by Sarepta to show an ability to promote greater dystrophin production in cells. The first is Exondys 51 (eteplirsen), an approved Duchenne treatment for patients amendable to exon 51 skipping.

An interim analysis of a Phase 1/2 trial was presented at the 2018 American Academy of Neurology (AAN) Annual Meeting in the United States. This clinical trial is the first to be carried out with humans and is a multicenter study, aimed to assess the safety, tolerability and efficacy of golodirsen in the target population, over a total period of 144 weeks or 2.7 years.

Exon skipping is a therapy that aims to restore adequate levels of dystrophin, the protein missing in boys with DMD. Dystrophin is essential to the structural integrity of muscle cells during contractions.

Golodirsen binds to the RNA sequence on either side of exon 53 and skips (excludes) this damaged part of a patient's genetic code for dystrophin and leads to the production of a partly functional dystrophin protein.

Escalating doses of golodirsen were tested against placebo to evaluate safety for the trial's first 12 weeks, then patients began receiving weekly treatment infusions at 30 mg/kg of weight. After 48 weeks of treatment, tissue biopsies showed significantly increased dystrophin production in all of the participants.

A Phase 3 clinical trial, called ESSENCE, is also evaluating golodirsen (SRP-4053) and SRP-4045's safety and efficacy in DMD patients with a genetic mutation amenable to exon 53 or 45 skipping, respectively.

A Belgian Blue cow that lacks Myostatin.

Therapy for FSHD Earns FDA's Fast Track Designation

Improving muscle strength and growth.

The U.S. Food and Drug Administration (FDA) recently granted fast track designation to Acceleron Pharma's ACE-083 for the treatment of facioscapulohumeral muscular dystrophy (FSHD). ACE-083 is a locally-acting agent that binds to and inhibits proteins, such as follistatin and myostatin, which are naturally occurring proteins in the body designed to regulate muscle strength and growth.

Though the first trials have been conducted with people affected by FSHD, ACE-083 may be useful for other neuromuscular conditions and the company now has a clinical trial

Continued on page 22

Continued from page 21

underway for Charcot Marie Tooth disease (CMT).

ACE-083 is designed to have a concentrated effect to maximise growth and strength in targeted muscles. Preliminary findings from Part 1 of the Phase 2 trial, which was an open-label, dose-escalation study of 36 patients with FSHD, demonstrated safety, tolerability and efficacy of ACE-083, with participants showing over 12% increases to mean total muscle volume in the lower legs (tibialis anterior) and upper arms (biceps brachii).

Participants received ACE-083 (150 mg or 200 mg) via intramuscular injection once every three weeks for 12 weeks. Total muscle volume changes were measured by MRI relative to baseline at 3 weeks after the last injection of ACE-083.

The company is now moving forward with Part 2 of the Phase 2 trial, which will be a randomised, double-blind, placebo-controlled study with open-label extension.

Fast Track designation by FDA means the company will be able to expedite the FDA review process for ACE-083, and if successful, this opens the door for marketing approval of the very first locally-acting 'Myostatin+' muscle agent as a meaningful treatment option for those affected with FSHD. Ⓡ

Gene Therapy Granted Orphan Drug Status for LGMD2E Treatment

Reversing the effects of the condition.

The U.S. Food and Drug Administration (FDA) granted orphan drug status to MYO-101, a gene therapy being developed by Myonexus Therapeutics for the treatment of limb girdle muscular dystrophy type 2E (LGMD2E). This is expected to provide additional regulatory support, financial benefits, and help expedite the clinical development and review of MYO-101 for its final approval.

LGMD2E is caused by mutations in the SGCB gene, which encodes the protein beta-sarcoglycan that is involved in maintaining the structure of muscle tissue. MYO-101 aims to permanently restore the levels of beta-sarcoglycan protein in the affected tissues, and reverse the effects of LGMD2E.

Mouse studies on MYO-101 have shown that a single intravenous injection of the gene therapy increased the levels of beta-sarcoglycan protein in all muscles. This was found to be accompanied by great improvements in muscle fiber size distribution and mean fiber diameter, and also decreased muscle scarring and fat infiltration. An 85.5 percent reduction of serum creatine kinase — a biomarker of muscle tissue deterioration — was sustained for six months following treatment. There were also several significant improvements in function, including increased diaphragm strength and resistance (required for normal breathing), reduced spine deformations, improved walking ability, and normalised cardiac output.

With such compelling preclinical data, The company is looking forward to initiating a systemic Phase 1/2a trial of MYO-101 later this year. Ⓡ

Positive results for Tideglusib in myotonic dystrophy

Improving the ability to perform daily tasks.

AMO Pharma have released interim results for a Phase 2a clinical trial of Tideglusib (also known as AMO-02) for adolescents and adults with congenital myotonic dystrophy. Interim findings suggest the drug is safe and well-tolerated, and may improve participants' cognition, fatigue and ability to perform daily tasks. The study findings also improved autism symptoms (where applicable).

AMO Pharma is developing the treatment for congenital myotonic dystrophy sometimes known as Steinert disease. Myotonic dystrophy type 1 (DM1) is an inherited condition that affects the muscles, heart, central nervous system, eyes, and endocrine system. Signs of congenital myotonic dystrophy typically show up at or soon after birth.

Diagnosis is done through medical evaluations or genetic tests. Preclinical-trial studies have linked DM1 to higher than normal activity of an enzyme called glycogen synthase kinase 3 beta (GSK3beta). AMO-02 inhibits the enzyme, improving muscle characteristics in tissue samples from DM1 patients.

The trial involved 16 people, aged 13 to 34 years, with congenital and juvenile-onset myotonic dystrophy type 1. Participants took daily doses of 400 mg or 1,000 mg of AMO-02 or a placebo.

Those on the 1,000-mg doses responded better than those on the 400-mg doses.

Both doctors and caregivers reported that the drug improved condition symptoms throughout the treatment period. The company is looking forward to further developing this clinical trial programme. Ⓡ

Omigapil shown to be safe in Phase 1 Trial for CMD

Congenital muscular dystrophies (CMDs) are inherited neuromuscular conditions characterised by congenital-onset weakness and hypotonia and have associated dystrophic findings on muscle biopsy. Progressive muscle weakness, joint contractures and respiratory insufficiency characterise most CMDs. LAMA2-related and COL6-related dystrophies are the most common forms of CMD for which no pharmacological therapy is currently available.

Santhera Pharmaceuticals have recently published findings on their preliminary investigation into omigapil, a potential therapy for CMD. In this single-center, interventional, open-label Phase 1 trial, researchers evaluated omigapil's properties, safety, and tolerability in 20 ambulatory and non-ambulatory patients between 5 and 16 years old with either Ullrich muscular dystrophy or congenital muscular dystrophy type 1A (MDC1A).

The ascending multiple dose cohort study (CALLISTO) met its primary objective, to demonstrate that the study drug was safe and well tolerated in children and adolescents with CMD. Participants were randomly divided into one of five groups and received omigapil once daily at a dose ranging from 0.02 mg/kg to 0.08 mg/kg body weight for three months. Data collected from the trial indicate omigapil is suitable for further development in the CMD population.

This was the first interventional trial with a drug candidate for CMD. Omigapil has orphan drug designations for CMD in the U.S. and Europe and was granted Fast Track Designation by the FDA. Further analysis in preparation for a pivotal trial will be done to provide evidence for drug marketing approval. Ⓡ

Limb-girdle muscular dystrophy

Understanding this group of inherited disorders.

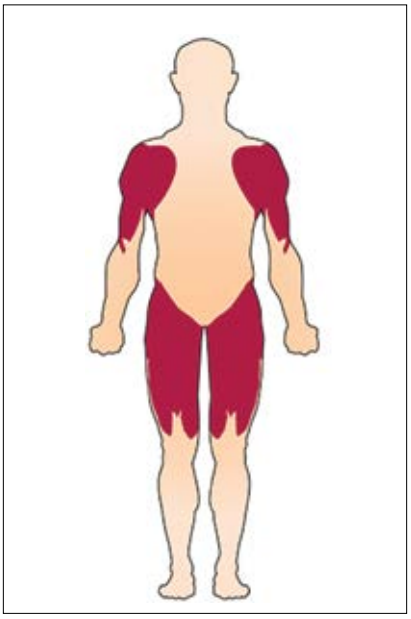
What is Limb-girdle muscular dystrophy?

Limb-girdle muscular dystrophy (LGMD) is a group of inherited disorders that cause weakness and wasting of the proximal skeletal muscles. These are the muscles closest to the body such as the hip and shoulder areas. The shoulder girdle is the bony structure that surrounds the shoulder area, and the pelvic girdle is the bony structure surrounding the hips. Collectively, these are called the limb girdles, and it is the observed weakness and atrophy (wasting) of the muscles connected to the limb girdles that has given this group of disorders its name. These conditions are progressive, and worsen over time.

In LGMD involuntary muscles of the digestive system, bowel and bladder are not affected, and sexual function is also normal. Intellectual and cognitive abilities also remain unaltered, as do sensations such as touch, temperature and pain.

The onset of LGMD varies and can occur in childhood or symptoms may not be apparent until adolescence or adulthood. Males and females are equally affected. Prevalence of LGMD is estimated to range from 1 in 14,500 to 1 in 123,000 individuals.

In the early stages of limb-girdle muscular dystrophy, affected



The muscles that experience weakness are highlighted.

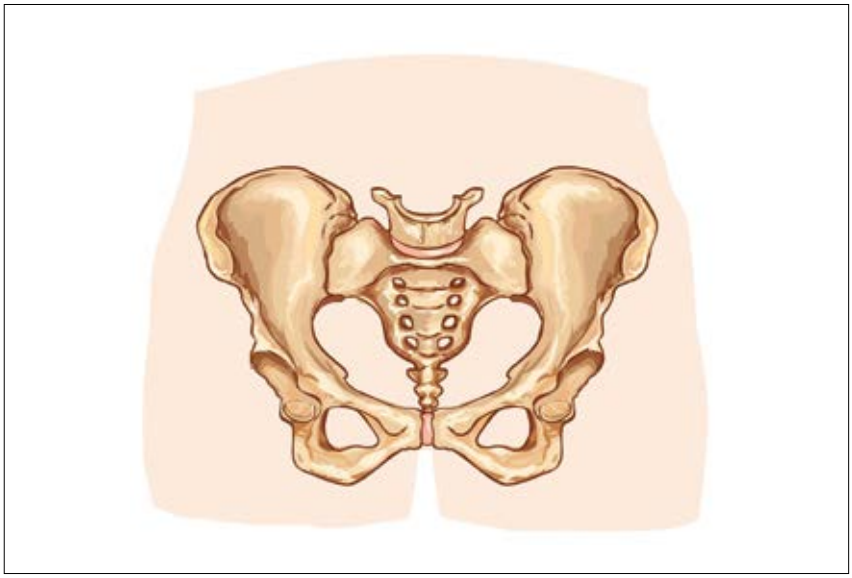
individuals may have weakness in hip and thigh muscles resulting in an unusual walking gait, such as waddling or walking on the balls of the feet, and may also have difficulty climbing stairs, running and getting up from a squatting or sitting position. The muscle weakness and atrophy may also result in lower back pain.

LGMD will progress to the shoulders, which can make reaching over the head, holding the arms outstretched or carrying heavy objects difficult. It may become increasingly hard to keep the arms above the head for such activities as combing hair. Some individuals find it increasingly harder to type, and may even have trouble feeding themselves.

Muscle wasting may also cause changes in posture. For example, weak shoulder muscles tend to make the shoulder blades protrude, known as scapular winging. Affected individuals may also have an abnormally curved lower back or a spine that curves to the side known as scoliosis. Spinal bracing may be required, and in more severe cases, spinal fusion surgery. An orthopaedic specialist is beneficial in monitoring scoliosis if present. Some individuals develop joint stiffness or contractures that can restrict movement in the hips, knees, ankles, or elbows. Surgery may be an option to release them. For some people, contractures may be an early sign.

Progressively, muscles of the face and distal muscles, such as the lower legs, feet, forearms and hands, may become affected and lead to considerable weakness. Calf muscles may appear unusually large (pseudo hypertrophy) as fatty deposits accumulate and replace lost muscle tissue.

Mobility may become increasingly restricted and 20-30 years from onset, individuals with LGMD may lose independent mobility and a wheelchair may be needed for mobility. Wheelchair options can be discussed with an occupational and/or seating therapist at the appropriate time.



The pelvic girdle is the bony structure surrounding the hips.

Genetics and classification of LGMD

Several different genes that normally lead to the production of muscle proteins have been identified as mutated in LGMD.

Most types of LGMD are inherited in an autosomal recessive manner, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Several rare forms of limb-girdle muscular dystrophy are inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

As there are many forms of LGMD, they are classified based on inheritance pattern and genetic cause, with '1' being designated for types that are inherited in an autosomal manner and '2' being the designation for those types which are inherited in an autosomal recessive fashion.

Mutations in the lamina gene (LMNA) cause LGMD1B.

LGMD1C is one of a group of muscle disorders called caveolinopathies caused by mutations in the caveolin gene (CAV3).

Calpainopathy, or LGMD 2A, is caused by mutations in the calpain (CAPN3) gene, this is the most common form, accounting for about 30 percent of cases. Dysferlinopathy, also called LGMD2B, is caused by mutations in the dysferlin (DYSF) gene.

Sarcoglycanopathies are forms of LGMD caused by mutations in the sarcoglycan genes (SGCA, SGCB, SGCG, and SGCD) These are known as types 2D, 2E, 2C, and 2F respectively.

A titan (TTN) gene mutation causes LGMD2J, which was first identified only in the Finnish population. Mutations in the ANO5 gene cause LGMD2L. Mutations in several other genes cause forms of LGMD called dystroglycanopathies, including LGMD 2I, 2K, 2M, and 2N.

Other rare forms of limb-girdle muscular dystrophy are caused by mutations in several other genes, some of which have not been identified.

According to the NZ NMD Registry 24 per cent of people have genetic confirmation of their LGMD.

As more and more genes are identified in the cause of LGMD, there will be a greater understanding of which and how proteins are implicated in the symptoms of LGMD.

Diagnosis usually commences after the identification of key early symptoms of LGMD.

Other rare forms of limb-girdle muscular dystrophy are caused by mutations in several other genes, some of which have not been identified.

Muscle biopsies show typical signs of damage, the presence of certain cell types, such as inflammatory cells, and can establish whether certain proteins are reduced or absent.

Electromyographies (EMG) can be used to observe the electrical activity of muscles and its consistency with activity typical of LGMD individuals. Blood testing can also be requested to look for elevated levels of creatine

A good diet with plenty of fresh fruit and vegetables is very important in ensuring excessive weight does not impede mobility.

phosphokinase (CPK) which are indicative of muscle problems. It is strongly recommended that genetic counselling is arranged. following a diagnosis of LGMD.

Implications and management

Cardiac problems can arise, such as weakness of the heart muscle (cardiomyopathy) or abnormal heartbeat (conduction abnormalities or arrhythmias). Arrhythmias can result in increased risk for heart palpitations (fast or irregular heartbeat) and syncope (loss of consciousness due to lack of oxygen to the brain). The heart must be monitored regularly and some problems may be controlled or treated with medication or devices (such as pacemakers), though severe forms can be fatal.

Respiratory muscles may also be affected resulting in breathing difficulties. When necessary, several options may be available to help maintain respiratory ability, ranging from exercises to the use of ventilators. Like cardiac problems, respiratory problems can be fatal and therefore need to be monitored closely. Many researchers have noted that progression of LGMD is often faster and more severe when the onset is earlier, in comparison to individuals who develop LGMD later in adolescence or adulthood.

From an early stage, it is important to undergo regular exercise and stretching programmes, with the help of a physiotherapist, to maintain muscle strength and flexibility. Swimming is an excellent option to exercise and mobilise all muscles and joints.

Combined with physical activity, a good diet with plenty of fresh fruit and vegetables is important to manage body weight, which helps with overall well-being and mobility.

Swimming is an excellent exercise option for people living with the condition.

On your bike

Andy Blay shares his journey with LGMD and the difference a healthy lifestyle – and a great bike – has made to living with his condition.

I was first roughly (and I mean very roughly!) diagnosed at the age of 19. My younger sister Carla was showing signs of fatigue and weakness, and Mum took her for a check-up. After a few tests they discovered her creatinine kinase levels (an enzyme measured in blood tests, usually elevated with injury or inflammation) were high, so they thought I better be tested too. As expected, my CKs were through the roof. After that, my sister and I had many different types of tests. To me, they felt like medical experiments at times.
The experts never really gave us a proper diagnosis, except for saying, "It's probably Limb Girdle Muscular Dystrophy". That was about it. I was told excess exercise would lead me into a wheelchair and to take it easy.

At the time, I was an apprentice carpenter and quite involved in sport including cycling and running. I really didn't know how to take the information I had been given. Should I stop being a builder? Should I stop exercising? I really didn't want to be in a wheelchair!! After contemplating this for what seemed like an eternity, I thought "Bugger it. I'm going to live my life like I had always planned to and what will be will be".

In the following years I did lots of exercise and finished my apprenticeship. I went on to do three Coast to Coasts and dozens of cycle races. I got married to a wonderful lady and we have two amazing children.

Unfortunately, as I aged my muscular dystrophy became progressively worse. I had to leave the building industry and got a job as sales rep for a furniture company. That job was perfect for me as it wasn't physically demanding. However, as I wasn't as active as I had been, I started to gain weight. As I put on weight, I noticed my MD got a lot worse. And I mean a lot worse! I couldn't believe how quickly I was deteriorating. I was drinking way too much (entertaining clients) and eating takeaways all the time. I couldn't walk properly, I was falling all the time, and my overall well-being was terrible. It got so bad I ended up using a wheelchair whenever I needed to be on my feet for more than 10 minutes at a time. Something had to change.

Fast forward lots of years and lots of tears (I'm now 42) and my life has changed a lot. My sister and I were finally diagnosed with LGMD type 2A. I have been a stay-at-home dad for nearly six years. My wife has a very busy job and it just made sense for me to stay at home with the kids. If I'm honest, my MD made the decision very easy as I was struggling to be on my feet all day. I am still walking (even if it isn't pretty) and I'm still very active, cycling whenever I can.

I find cycling the perfect activity for me as its non load-bearing and

Stay-at-home dad Andy loves cycling with his children.

on a bike I feel like a normal person, not the stay-at-home dad waddling into school to pick up his kids. I have a very simple approach to managing my MD. I believe the healthier I am, the less my MD affects me. I am now at a healthy weight and try to treat my body the best way I can. It is my personal belief that regular exercise and a good wholefood diet key to slowing the progression of this illness.

While it may not work for everyone, its certainly working for me. I feel better than I have in years, and have lots of energy to tackle my busy days. I'm now volunteering for a programme at Cycling Southland called Cyclofit. We use cycling as a form of rehabilitation, and general fitness for a variety of people. I get a huge sense of satisfaction helping people with their fitness and overall health. Cycling is an excellent way to achieve that.

Let's meet for coffee

NATALIE BRUNZEL

That simple suggestion requires a lot of preparation, says Natalie Brunzel.

When a good-looking guy came up to me and suggested. "Let's meet for a coffee," instead of excitement, a feeling of dread came over me. Why? Coffee might sound like a stress-free activity, but when you use a power chair, that simple request brings to mind an array of logistics to be worked through.

Over the years I have found ways to make meeting for coffee an easier experience. Cafés are social hubs these days. They are places for social interaction, but also double as an office at times. Ironically, I have had an interview in a café when the office wasn't wheelchair accessible. Although they serve multiple purposes, it comes as a surprise at how inaccessible many cafes are.

One of the best tips I've been given is to order my drink in a takeaway cup.

Once the day and time of our coffee date has been decided, the next step is choosing the café. For me, a café needs five main ingredients to work well. One; there must be mobility parking. Two; I must be able to get inside and up to the counter without any obstructions. This sounds like easy, but you would be surprised how many places have their tables packed so closely together I can't get past. Three; my wheelchair needs to fit under the table. Often there is nowhere for my footplates to go, and I am stuck jutting out in the way of all the other customers. Four; potentially one of the most important points, the café needs an accessible way to pay. I'd hate to have to count up the number of times my friends have had to pay for me because the cable for the eftpos machine wasn't long enough to reach over the counter, and not everywhere has paywave. The way to get around this embarrassment is to either carry enough cash to settle the bill the old-fashioned way, or check out the payment options beforehand.

The final thing I look for in a café is the attitude of the staff. If they are friendly and helpful, I feel confident enough to ask them for help should an awkward situation arise

After all the logistics have been scoped, I can finally allow myself to feel excited about a coffee date. Then there is just enough time to try on five different outfits, sending snapchats to my bestie asking for advice, before heading out to meet my date.

One of the best tips I've been given is to order my drink in a takeaway cup. Many café cups are just too heavy or difficult for me to hold, and this cuts down the anxiety about spilling a hot drink – I can leave that job to the waitress!

Natalie Brunzel works as a freelance Communications Consultant. She has worked within the disability sector for more than 10 years. Her extensive knowledge of the disability sector, and lived experience of an impairment provides a refreshing look at topical issues.

All about me

SCOTT BOYLE

Introducing our new Rangatahi Representative on National Council, who shares about silencing his inner critic.

Allow me to properly introduce myself. My name is Scott Boyle and I am the new Rangatahi Representative for MDANZ. Naturally, you won't know who I am as I've only recently become involved in the organisation. To summarise, I am 23, have Spinal Muscular Atrophy type 2, love to have a good laugh and mess with people, and live in the rather uneven and cracked city of Christchurch. One of the most defining aspects of who I am is that I'm a writer. For the past year I have been working on a series of novels. Writing and telling stories are things I have enjoyed since I was very young, and eventually people began encouraging me to follow my passion. And thankfully they did, for I am growing into a rather ambitious storyteller!

I'd like to share a little bit about why I became involved with the Muscular Dystrophy Association.

In October of last year I almost died after catching the flu, which quickly progressed into pneumonia. It was the first time in years that I had truly tasted that fear of death again. But after I pulled through and began the journey to recovery, I felt the need for change. For three years I had put my life on hold whilst attempting to cope with chronic pain, and that had robbed me of time I could have put to better use. So by the New Year I was back to full strength and ready to seize whatever opportunities came my way. That's when the universe sent me the fabulous fieldworker Paul Graham. Paul told me about the vacancies on the Canterbury Branch and encouraged me to join.

When he realised how genuinely enthusiastic I was he told me about the role of Rangatahi Representative becoming vacant on National Council, with Olivia Shivas standing down after 2 years. That night I thought a lot about whether or not to put my name forward. What sparked my hesitation was a little whisper in the back of my mind that stated; "You have never done anything like this before, what right do you have to be a candidate?" And a part of me knew the voice was right.

We all have those moments. A lot of the time they are enough to hold us back. Other times however, we can silence that bothersome voice and grab the reins to steer our own lives. What convinced me to do so was the notion that I might be able to help even one person in our community, and that I could have the pleasure of meeting more of you amazing people. So here I am MDANZ!

Hanging out with a new friend.

We all have those moments. A lot of the time they are enough to hold us back. Other times however, we can silence that bothersome voice and grab the reins to steer our own lives.

Scott Boyle lives in Christchurch and is the Rangatahi Representative on National Council. He loves history and storytelling, and is currently working on his first novel. He is passionate about raising awareness of muscular dystrophy and ensuring younger voices are heard.

Ask the **PANEL**

What's in a diagnosis?

DR. RICHARD ROXBURGH

Q: How can you have a diagnosis, but still be "undiagnosed"?

Last week I attended the NZ National Huntington's disease (HD) conference here in Auckland. I've attended these conferences regularly over the last 15 years and previously, to be honest, I've come away fairly sober if not in tears at the families' stories. But this time there was something different. Twenty-five years after the discovery of the gene for HD there is clearly hope that disease modifying treatments are just about here.

During the course of the conference I had a conversation about my passion for finding new genes – and it occurred to me how much a difference there is for those who have a genetic "diagnosis" and those who are still wandering in the wilderness without.

How can there be this difference? And what does it mean when you have a "diagnosis" but not a genetic diagnosis. It mainly comes down to how distinct a condition is. Huntington's is a condition which is very distinctive and so when someone presents with this characteristic pattern of symptoms it is relatively easy to deduce that they have the genetic abnormality. In turn that has led to (relatively) rapid understanding of the disease – it also helps that it's not too rare.

The other situation is when a whole lot of different diseases caused by different genes all present with the same clinical pattern. In such cases the patient may be given a "diagnosis" which is more a description of their condition. One example would be "limb girdle muscular dystrophy" – literally a "disease of the muscle around the shoulders and pelvis" or "spinocerebellar ataxia" – literally a "disease of the spinal cord and the cerebellum which causes incoordination". These too are "diagnoses" but if we don't know the underlying genetic cause (e.g. limb girdle muscular dystrophy type 2a caused by mutations in the calpain gene) then they don't have such power to tap into the research and thereby the hope of the transforming treatment possibilities that we are beginning to see in genetic diseases and which is beginning to bring hope at Huntington's conferences.

In the last five years, with "Next Generation Sequencing" there has been a huge transformation in our ability to give specific genetic diagnoses to patients. However, the tests don't diagnose everything because we still only know a fraction of what there is to know about the human DNA code, e.g. even in the best labs the diagnosis rate for limb girdle muscular dystrophy is still only 45 percent. Patients who test negative are still left with the descriptive diagnosis.

We need two things 1) a catch up programme for the hundreds of people in New Zealand who have conditions that can now be diagnosed at the genetic level but haven't been, such as some of the 76 percent of patients with limb girdle muscular dystrophy in NZ who don't have a genetic diagnosis. And 2) ongoing research to find those new genes.

There is a Catch-22 situation here. There is a lot of research money available for finding new genes but a reluctance to provide research money to screen patients for known genes; at the same time there is a reluctance to spend clinical money on genetic tests whose main motivation is to allow the person to take part in future, as yet unspecified, research studies. This is a conundrum that I've been wrestling with for some time now and we will continue to work on it.

Dr. Richard Roxburgh FRACP PhD is a Consultant Neurologist at Auckland Hospital and Associate Professor at the University of Auckland's Faculty of Medical and Health Science.

Managing pain

MIRIAM HANNA

Living with a neuromuscular condition often means living with pain.

Pain can be acute, coming on quickly and resolved within hours or days, and can be a result of injury or external insult to the body, incorrect positioning or strain or sprain. Chronic pain on the other hand, is long-term pain, and can be harder to manage.

It is important to manage pain effectively as it affects many aspects of life. Pain affects sleep, which in turn affects the ability to carry out daily tasks. It may also affect appetite, mood, participation in family, work and social life, and may lead to depression or anxiety.

Improving pain management and alleviating pain results in a significantly improved quality of life. It is important to discuss pain with your doctor and, if possible, understand the root cause of it. This will help identify best options for management.

There are two distinct types of pain, musculoskeletal and neuropathic. Musculoskeletal pain is much more prevalent, and is pain resulting from injury to muscles and bones. This can include strains, sprains, fractures, cuts and bruises, mostly acute types of pain. In some cases however, musculoskeletal pain can be chronic. Treatments for musculoskeletal pain include analgesics such as paracetamol and tramadol, or anti-inflammatories such as ibuprofen and diclofenac.

Neuropathic pain results from injury or disease affecting nerves. People with neuropathic pain often experience very different sensations to musculoskeletal pain, including burning, stabbing, shooting or electric sensations. This can be constant, or in response to triggers. Long-term neuropathic pain can cause central sensitisation, resulting in exaggerated pain in response to mildly painful stimuli (hyperalgesia) or pain in response to innocuous stimuli (allodynia). Neuropathic pain can be central and caused by brain related disorders such as multiple sclerosis, stroke or traumatic brain injury, or peripheral, caused by cancers and chemotherapy, diabetes, hereditary neuropathies (like CMT), radiculopathies, surgery or amputation or trauma.

Treatment of neuropathic pain with medication is tricky. People are often on numerous medications, including opioids, tricyclic antidepressants or antiepileptics, which are not primarily indicated for neuropathic pain treatment, but used in some cases to reduce the pain. We all respond differently to these medications, and what might work for one person might not work for another. Doctors may use a variety to methods to examine or diagnose neuropathic pain, including sensation testing, nerve conduction studies, CT scans or MRIs, genetic testing, skin or nerve biopsies, questionnaires, blood tests, and individual history or pain diaries.

Pregabalin is a funded treatment available specifically for neuropathic pain. If you suffer long-term neuropathic pain, discuss trialing pregabalin with your doctor.

Non-pharmacological approaches to reducing pain include improving sleep by keeping to a regular routine and reducing caffeine, alcohol and the use of electronic devices in the evening. If your activity level is low, a green prescription for exercise may be beneficial. Some people find massage, stretching, osteopathy and meditation, are helpful to manage pain symptoms.

Topical treatment of localised neuropathic pain with creams is also an option. These include anesthetics as lidocaine and prilocaine or capsaicin.

MDANZ has a limited number of RUBEEVEN samples provided by HoneyLab Ltd. This is a dual action product available in pharmacies, containing capsaicin which produces a topical warming effect, and bee venom for joint comfort. If you would like to trial the product please email info@mda.org.nz and quote your membership number. Please note this is not right for you if you are allergic to bee stings.

Miriam Hanna is Member Services Manager 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
Member Services Manager

Melanie Kerr
Executive Assistant

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

Northern Branch


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

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

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