Winter 2021

MNDnews

The newsletter of the Motor Neurone Disease Association of Tasmania

RESEARCHERS CELEBRATE MND WEEK

mnd Tasmania

Until there's a cure, there's care

MESSAGE FROM THE PRESIDENT

Our AGM in March every year is a highlight on our calendar, and this year was no exception! The meeting in 2020 was held in Devonport on the threshold of a pandemic. This year, we met in Hobart at the Howrah Community Centre on 21st March.

The AGM provides us with the opportunity to be accountable, meet new members, review the past year, and hear from a guest speaker. Associate Professor Bradley Turner is the Head of the Motor Neurone Disease Laboratory at the Florey Institute of Neuroscience and Mental Health, Victoria. We were privileged that he attended our meeting and presented his team's interesting and ground-breaking research on "Developing personalised medicine approaches for MND" clearly, and at a level that we could understand. We were also fortunate to have Eric Kelly join us on behalf of MND Victoria. He is the Team Leader of the MND Advisor and NDIS Support Coordination Service in Tasmania.

We will embark on this new year with the addition of three new board members, supported by the remaining team with continuity of experience and knowledge. Changes to the Constitution were successfully adopted and the structure of the Board has changed accordingly.

After being our President for two years, Michelle Macpherson has retired so that she can return to her core role of Fundraising Coordination. I have been honoured to be able to accept the role of President because Lucy Polizzi has joined us as Secretary. We are pleased to have the portfolio of IT filled again, and Steve Isaac has taken on this task as we plan to change the MND Tasmania website, upgrade document storage, and improve our social media content. Jules Driessen is continuing as Treasurer, and we are pleased that Rendell Ridge continues to offer his time pro bono to audit our finances year by year. Chris Symonds will continue as Public Officer and Elisa Howlett is joining us to take over from Libby Cohen in coordinating member supports. Professor Tracey Dickson, who filled a casual vacancy during 2020, was also elected, and will take on the role of representing MND Tasmania on the Board of MND Australia.

At every AGM, we formally remember those who have passed away due to MND in the past year. On behalf of MND Tasmania, I extend our heartfelt condolences to the families and friends of those who have recently passed away.

Please write and share your stories, opinions, questions or any thoughts about this newsletter. Write to me at info@mndatas.asn.au and see our contact details on the back page.

Kate Todd
President

MND TASMANIA SAYS **THANK YOU**

Thank you Michelle

We would love to say a big thank you to Michelle Macpherson for all of her hard work and dedication as President of MND Tasmania for the past two years. Michelle has stepped down from this role, but we are delighted that she will be staying on in her core role as Fundraising Coordinator.

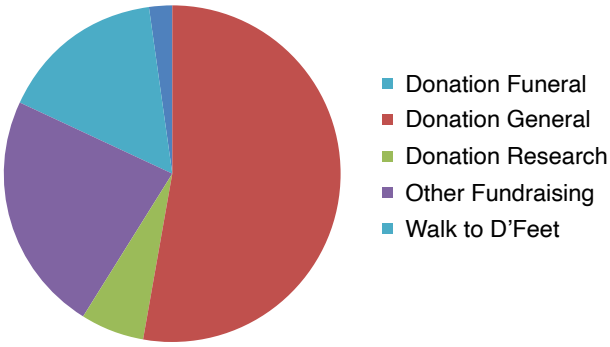
Thank you to our community

As with many organisations, our income from donations was heavily impacted in 2020 with the cancellation of and/or absence of many fundraising events due to COVID-19.

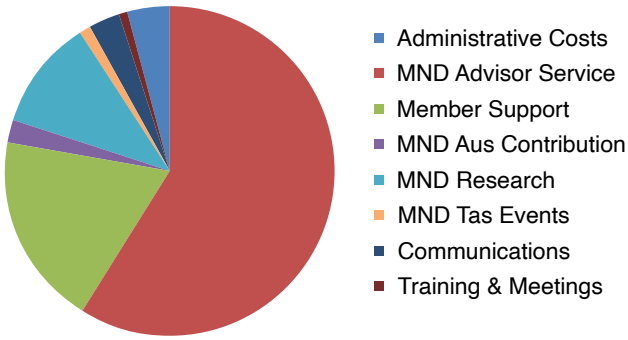
We were, however, still fortunate to receive a number of generous donations from our community. With these funds, we were able to continue to contribute to the operation of the MND Advisor service in Tasmania, provide member support by way of equipment and home modifications, and donate to MND research.

Every single contribution made to MND Tasmania, regardless of its size, helps to make a difference in the lives of people living with MND. So we say a big thank you to you, our MND Tasmania community, for your continued generosity and support.

Income Financial Year End 2020



Expenditure 2020



Researchers Celebrate MND Week

Our MND Researchers at Menzies Research Centre, Hobart, aren't only world-class scientists, they can also bake up a storm!

They got together to mark MND Week 2021 and to take a well-earned morning tea break. MND Week is an opportunity to raise awareness of motor neurone disease, highlight the importance of advocacy and best practice care, and promote the need for continued research to end MND. We will continue to work to understand the cause of this terrible disease and to develop new treatments to stop it in its tracks. But until that day, MND Tasmania will be here providing the best possible care and support for all Tasmanians living with MND now and in the future. Because until there's a cure, there's care.

Thank you Joyce and Libby

Our Association is managed by a Board of volunteers and has been incredibly fortunate to have had the commitment and experience of our two longest serving Board members, Joyce Schuringa and Libby Cohen with us for so long.

Their contribution has been significant, and we were sad to farewell them as they retired from the Board. We wish them the best of luck in all of their future endeavours.

LIVING BETTER

Communication and MND

Communication is an integral part of our daily lives. Communication involves more than communicating by speech and using our voice.

Social communication is how we interact and engage with the people in our lives and the broader community. If you are experiencing communication changes due to MND, it is helpful to know that there are lots of strategies and supports available.

Here are some practical tips to help you communicate:

- **Be mindful of the environment** – reduce noise, face your communication partner, allow yourself extra time.
- **Consider using technology to support you to communicate a different way** – this may be using a text to speech app (on mobile phone or tablet) or using a communication software with an eye gaze device.
- **Have other ways of communicating beyond talking** – using a pen/paper, pointing to a photo on a board, using gesture, or responding to yes/no questions.
- **Conserve your energy to support your communication abilities.** Some examples include – resting before social events to enable you to communicate more easily; or connect with your friends/family through text messages rather than phone calls if you find this easier.
- **Pictures are worth a thousand words!** Use photos to enhance your social communication. Take photos of activities that you do throughout the day. Send the photos to a family member, friend or community group. Ask that they send you photos back. See how this has a positive impact on your social interaction!
- **Use social media (e.g. WhatsApp, Facebook Messenger) to create private groups.** Private groups are a great way to keep in touch with family, friends or other community groups you are involved in (e.g. cricket club, Rotary club, etc.). It allows you to easily send messages and/or photos so that you can stay socially engaged with the people in your life.
- **Spend some time in nature.** Nature has been shown to positively contribute to wellbeing and provides an opportunity to slow down, reflect, and create a 'mindful moment'. You may feel inspired to take a photo of something that you find beautiful or that catches your eye. Share this photo with the people in your life. You may like to create an Instagram page, public or private Facebook group, or send it around to your family or friends.
- **Send a daily or weekly email around to the people in your life.** Like a newsletter, include a summary of things that you find funny, quotes, photos, or anything! Let your personality shine through.

A speech pathologist can support you to communicate as effectively as possible with the people in your life. We will listen to what is important to you, who are the meaningful people in your life, and assist you to come up with ways to communicate and stay socially connected with the people who matter to you. An occupational therapist may also be involved to explore new ways of accessing your communication equipment or technology if your hand/arm function is changing.

Kaela Gomizel
Speech Pathologist,
Calvary Health Care Bethlehem

PERSONAL STORY

Keeping Our Eyes on the Stars

by William Pridmore (MChD) Royal Hobart Hospital Hobart, Tasmania, Australia

Originally published in RRNMF Neuromuscular Journal 2021;2(1):8-9

The real problem is time.

She was finding it a bit harder to lift her foot. It was irritating, and she found she was limping. She made passing comments to her husband and son of some "electrical shocks" in her legs, which she thought were due to anxiety. That combination worried me, but I didn't tell her. It couldn't be.

A "foot drop" is a symptom in which the muscles that lift the toes towards the head lose this ability. There are many causes for such a presentation, and determining the culprit is challenging. Things didn't improve, and so she underwent a series of investigations. The neurologists who administered the nerve conduction studies were kind, but she noticed they exchanged serious looks throughout the test. She'd have to wait for the full report.

The full report was not good.

When the diagnosis of motor neurone disease is made in a family member, the world changes. It darkens around the edges, and loses some of its lustre. One can deal with the impending physical challenges as they arise. But it's the time that gets you. For most patients, lifespan is between two and five years from diagnosis. Nothing stops the march towards oblivion. We know how this story goes.

Massachusetts native Dr Stanley Appel is one of the rock stars of this condition. At 87, he valiantly continues the research and clinical work he began as a young doctor. He calls MND "nice guys' disease", based on his observations that MND patients are typically driven, empathetic, generous, and otherwise healthy. Scientific studies, comparing patients with a healthy control population, confirm his notion [1]. MND patients are card-carrying nice guys – that's part of the injustice.

Research into MND has been frustratingly slow since the condition was first described by French neurologist Jean-Martin Charcot in the 1860s. Degeneration and death of motor neurones (nerve cells that control muscle movement) in the brain and spinal cord leads to progressive paralysis of voluntary muscles. The causes have remained elusive, and the biological processes that go astray are highly complex.

Most cases occur at random, but a minority run in families. When the first known genetic contributor was identified in the 1990s, it inspired the first animal model of MND, using laboratory mice. Such mouse models of MND have provided important insights about the injury of motor neurones, and have offered a useful way to test for the beneficial effects of various drugs. Unfortunately, these models have failed to bear much fruit. Contributing factors include poor experiment design, incompatibilities between mouse and human disease, and inadequate understanding to allow for informed therapeutic proposals [2].

My name is Will, and I was thrust into the MND community after my mother, Mary, was diagnosed in June last year. I am a doctor by trade and wrote this article to share the impact of this diagnosis on a family, as well as to provide some context and optimism regarding the hunt for treatments. We are grateful for the support provided by MND Tasmania as we navigate this condition.

But something is shifting.

Decades of work by devoted researchers have discovered much of what leads to MND. There is more optimism than ever before. Slowly but steadily, scientists are developing a working knowledge of motor neurone death. In the same way MND insidiously takes over a body, research is progressively uncovering its secrets – and will eventually banish it. Several drugs and treatments show early promise.

2020 saw the launch of three MND "platform trials": one in the UK, one in the USA, and one in Europe. Regularly used in cancer research, platform trials allow several drug candidates to be compared to a single placebo (fake drug) group at the same time. Contrasted with traditional clinical trials, platform trials may cut testing time in half, and expense by a third. The arrangement also means fewer patients who participate in trials will receive placebo. For a platform trial to be viable, there must be enough medications in development. For a drug company to develop a medication, it must know enough about a particular condition to identify a likely therapeutic target. Drug company interest in MND has increased out of sight over the last five years [3].

University of Michigan linguist John M. Lawler offers the term "railroad time". It describes how, when science and technology is sufficiently advanced, it is natural for several people to make a discovery concurrently. Railroads were invented when it was "time for the railroads". I guess it is "railroad time" for MND platform trials.

Key discoveries hint that laboratory tests to reliably diagnose and monitor nerve damage may soon be available [4]. "Biomarkers" like these are desperately lacking. Implementation of such tests would revolutionise drug development, as researchers could tell quite quickly whether their medication was having an effect. The code is being cracked. You can hear the pins falling into place.

For now, we have learned to celebrate the small things, and seek joy. A new orthotic has stabilised her walking. Her arms are still strong – suddenly her love for kayaking is amplified. Her painting studio, always a special place, is now her salvation.

It is clear that MND is increasing in prevalence [5]. Despite its dramatic effects, the condition remains relatively underfunded. The Fight MND Foundation, championed by Australian Football League great Neale Daniher, has brought needed attention to our plight – and other excellent organisations around the world continue to raise funds.

More is needed, and needed quickly. Two Australians are diagnosed with, and die from, MND every day. The battle against this condition has felt like Sisyphus eternally pushing his boulder up a hill, only for it to roll back down when it nears the top. But now there is a difference. With renewed awareness and financial support, we may actually, finally, reach the summit.

And hopefully Mum will be standing with us, waving the flag of victory.

References

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4. Benatar M, Zhang L, Wang L, et al. Validation of serum neurofilaments as prognostic and potential pharmacodynamic biomarkers for ALS. Neurology 2020;95(1):e59-e69. DOI: 10.1212/ WNL.0000000000009559

5. Longinetti E, Fang F. Epidemiology of amyotrophic lateral sclerosis: an update of recent literature. Curr Opin Neurol 2019;32(5):771-776. DOI: 10.1097/WCO.0000000000000730

MND RESEARCH

Unravelling the genetic origins of MND

While the understanding and awareness of MND continue to grow, the underlying causes remain largely unknown. To date, the only proven cause of MND is gene mutations that are essentially typos in the letters of our DNA sequence.

Clinicians first described MND in the mid-1800s and, soon after, a large family from Vermont was identified in which multiple individuals were affected by MND. This family provided the first clue that there is a hereditary or genetic component of MND, and we now know that approximately 10% of all individuals affected by MND have a similar family history, and are classified as having familial MND. Though clinicians gradually expanded the understanding of the clinical presentation of MND, the first major breakthroughs in understanding the biology of MND did not come until the 1990s. It took 113 years from the identification of the Vermont family, to finally solving their cause of MND as a genetic mutation within the SOD1 gene, representing the very first MND gene discovery in 1993. In 2006, the principal component of protein clumps apparent in MND was found to be a protein called TDP-43. This led to genetic investigations of the gene encoding this protein, TARDBP, which in 2008 revealed that mutations within this gene do, in fact, cause MND. These discoveries demonstrated the crucial interplay of the genetics and biology of MND, and researchers have continued to use genetics to inform the further study of the biological mechanisms involved in MND.

In 2009, a powerful DNA sequencing technology called next-generation sequencing was developed, which can produce enough genetic data to provide a complete genomic profile of an individual within just a few days. This technology has facilitated an explosion in the number of MND genes discovered over the past decade, both by our Australian research team and by geneticists across the world. However, these known MND genes still only explain two-thirds of families afflicted with MND, and ~7% of individuals affected by the non-familial or sporadic form of MND, leaving up to 90% of all people affected by MND without any identified genetic predisposition to MND. However, it has been shown that ~50% of the risk for any form of MND, whether familial or sporadic, is attributable to inherited factors. Therefore, the identity of many of the genetic changes contributing to the cause of MND remains to be discovered.

An important theory has been postulated by our collaborators in the UK stating that six individual events or "hits" need to accumulate within an individual to trigger the onset of MND. These hits could be genetic risk factors or environmental exposures we are yet to identify. Supporting this theory, our research team has recently shown that among people affected by MND, those who carry multiple MND mutations and/or genetic risk factors develop MND earlier in life than those without any such genetic alterations.

Our research team has collected DNA samples from over 600 Australians affected by MND and have utilised next-generation sequencing to determine the complete genetic profiles of each of these individuals. This is an incredibly powerful resource containing a huge amount of genetic information. In addition to analysing families affected by MND using approaches similar to those used to identify SOD1 as the first MND gene, we are also looking for MND genetic risk factors, which are genetic alterations that, while present in the general population, are more common among people affected by MND. However, as these genetic risk factors only contribute to small levels of MND risk, they are very difficult to identify, and very large numbers of study participants are required for their discovery. Our team is involved in a large international consortium called Project MinE, which collects genetic profiles from individuals affected by MND from across the world. So far, over 10,00 genetic profiles have been collected for analysis in a coordinated effort to discover novel genetic risk factors for MND.

MND genes are used for diagnostic testing and facilitate early diagnosis of MND, which allows clinicians to provide efficient management strategies with a view to delaying progression. This affords family members opportunities to utilise genetic testing if they so wish, under the guidance of skilled genetic counsellors. This includes pre-symptomatic testing to understand their own risk of developing MND, and/or the ability to utilise preimplantation genetic diagnosis together with IVF to prevent future generations from inheriting MND-causing mutations. MND genes and the proteins they encode are also prime targets for developing effective therapeutic interventions to treat MND. While not yet a reality, we are working towards using genetic profiling to tailor treatment strategies for each individual. Together, these benefits of MND gene discoveries are driving us toward personalised medicine approaches to treating MND within individuals and families in the hope that we can ultimately outsmart and beat the beast that is MND.

Emily McCann, PhD
Beryl Bayley MND Postdoctoral Research Fellow
Centre for Motor Neuron Disease Research,
Macquarie University

UPCOMING EVENTS

10th National MND Australia Conference

The 10th National MND Australia Conference is scheduled to be held online on Friday 3rd September 2021.

The National MND Australia conference is for clinicians, health professionals and disability and aged care providers from across Australia and NZ who have an interest in, and care for, people living with MND. People living with MND, their families and friends are also encouraged to register. Attendees will be updated with evidence-based clinical and practical information, and will also have the opportunity to network with fellow practitioners, share expertise and problem solve.

The Conference aims to promote sharing of expertise and understanding of MND nationally to improve care and support for people living with MND, their carers, and families. It provides an opportunity to promote evidence-based and best practice care, and to encourage collaboration, coordination, research and a multidisciplinary team approach.

For more details and to register, please visit https://www.mndaust.asn.au/conference.aspx or scan the QR code to the right.

Northern MND Support Group

This is an informal coffee and catch-up style group, where members come together to talk all things MND and Kennedy's Disease, as well as share experiences and helpful tips.

WHEN: Every second Tuesday at 10 am

WHERE: Selah Café at the Door of Hope, 50 Glen Dhu Street, South Launceston

CONTACT: Deb Beyer is the coordinator and can be contacted on 0418 462 369, 1800 777 175 (message) or email: info@mndatas.asn.au

North West MND Support Group

The NWSG in Ulverstone invites all MND Tasmania members and supporters to its meetings.

WHEN: At 11 am on the first Wednesday in the month, except January

WHERE: Ulverstone Returned Servicemen's Club Back Room, 21 King Edward Street, Ulverstone

CONTACT: Junene Stephens is the secretary and can be contacted on 1800 777 175 (message) or email: info@mndatas.asn.au

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GIVING

Port Sorell Primary School Ice Bucket Challenge

Many thanks to the staff and students at Port Sorell Primary School who, in March, dressed up in blue and enjoyed a BBQ lunch, and then every staff member donated $10 to enter the Ice Bucket Challenge. This special event was to support a colleague who is battling MND. Many thanks from MND Tasmania for your amazing efforts!

WHY GIVE

MND Tasmania is a volunteer organisation receiving no government funding. We are dependent on the generosity of our community and would like to acknowledge and sincerely thank our donors, including Port Sorell Primary School and Hearts and Crafts in Perth, Northern Tasmania.

We have also gratefully received donations in memory of Alain DeSousa.

Every single contribution made to MND Tasmania, regardless of its size, helps make a difference in the lives of people living with MND.

To show your support, visit https://donate.mycause.com.au/charity/6483 or scan the QR code below.

MND ADVISORS AND NDIS SUPPORT COORDINATORS

Client Numbers

We are currently supporting 44 registered members:

North West Region Tasmania 8
North Region Tasmania 20
Southern Tasmania 16
TOTAL 44

CURENTLY SUPPORTING
44
REGISTERED MEMBERS

DELIVERED
654
HOURS OF SERVICE

Above: Emma Forsyth
Left: Jenny Fuller

Board Members:

MND Tasmania has a volunteer Board and no paid staff.

President: Kate Todd

Senior Vice President and Fundraising: Michelle Macpherson

Vice President & Public Officer: Chris Symonds

Secretary: Lucy Polizzi

Treasurer: Julie Driessen

Member Support: Elisa Howlett

Ordinary Directors: Tracey Dickson, Steve Issac

Ex-officio Support: Pam Steele-Wareham, Libby Cohen

MND Advisors and NDIS Support Co-ordinators

Northern Tasmania: Emma Forsyth (Mon, Wed, Thurs):
p: 0456 182 551 or e: eforsyth@mnd.asn.au
Emma has moved to new office space at Launceston General Hospital.

Southern Tasmania: Jenny Fuller (Tue, Wed, Fri):
p: 0412 599 365 or e: jfuller@mnd.asn.au

Team leader: Eric Kelly (Mon-Fri.):
p: 0421 323 850 or e: ekelly@mnd.asn.au

FREECALL 1800 777 175

This number is at the MND Victoria office (Monday - Friday, 9 am - 5 pm). To assist the volunteer receptionist, please say that you are calling MND Tasmania. If you wish to speak to a Board member or an MND Advisor, you will be asked for your contact details and your call will be returned by that person as soon as possible.

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