

talking point

NEWSLETTER OF MUSCULAR DYSTROPHY NEW SOUTH WALES

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Camp Kula N' Gadu



**MUSCULAR DYSTROPHY
NEW SOUTH WALES**

A Word from the President

A few weeks ago, I saw the first Christmas messages rolling out in shops and it jolted me into realising 2013 is drawing to a close. While the year has been challenging, it's also been a good year for your Association and based on the feedback we're getting, next year should be even better.

One of our excellent services is the Camp at Narrabeen. We've just completed our 2013 Camp where we welcomed The Honourable John Ajaka MLC, our new Minister for Disability Services, who came along to meet our campers, carers and supporters. The Minister was obviously moved by the sheer delight of our campers and the wonderful care and dedication of our staff and volunteers. I wasn't surprised by the Minister's reaction because, to my mind, our Camps really do represent a window on the success of MDNSW in providing invaluable and ongoing support to our clients and their families.

With the Federal Election now thankfully behind us all, we now stand on the threshold of the new era designed to really empower Australians affected by disabilities. At MDNSW, we are proud to be playing our part in supporting the introduction of these changes. We are rapidly moving into the new era of change with the National Disability Insurance Trial well underway.

But our Association's role remains clear - MDNSW is the leading whole of life provider of choice of specialised services and expert information for people affected by neuromuscular conditions. And to keep our edge, our Association must continue to evolve as we seek to deliver the best possible services for you.

As I've mentioned before, we do need your help. Regular advice and feedback are essential if we are to remain a Centre

of Excellence for Muscular Dystrophy Services. Please take every opportunity to give us feedback - good or bad - whenever it crosses your mind. Brief emails to our CEO Pene Hodge (pene.hodge@mdnsw.org.au) are the best way for us to get feedback.



Also don't forget there are many people in New South Wales with neuromuscular conditions we are yet to reach. If you are aware of someone with a neuromuscular condition and you know they have not been in contact with MDNSW as yet, please give them a copy of Talking Point and suggest they give us a call. Tell them that MDNSW and our supporters are ready to help and we want to reach out to them.

I must add that our work is always greatly assisted by those visionary people who make our Association a beneficiary in their Will. Their vision and generosity allows us to reach out to more and more families across the state with better services. If you are about to update your Will, please consider including The Muscular Dystrophy Association of NSW as a beneficiary so one of your legacies will be underwriting our work into future years.

And finally, thank you to one and all in our Muscular Dystrophy NSW family for all your efforts.

Peter Debnam

President, Muscular Dystrophy New South Wales

Good Luck to everyone participating in our Big Red Ride in December!

Registrations are officially closed however if you would like to donate to a rider please go to www.bigredridesydney.com.au



Sunday December 1



**MUSCULAR DYSTROPHY
NEW SOUTH WALES**

A Word from the CEO

I have been sitting in my office contemplating the wonderful week we have just had at our Camp Kula N' Gadu where forty children and young adults from throughout NSW had the time of their life. The meaning of Kula N' Gadu is to be proud of who you are, to be strong, to discover all that you can be and to shine, and our aim is to ensure that every camper has the opportunity to do this.

“ The Camp is an excellent example of how our Muscular Dystrophy NSW community works together. ”

We couldn't do it without the support of ADHC, the doctors and therapists from the Neuromuscular Clinics, the occupational therapy students from UWS and the very many volunteers who year after year give their time.

At this year's Camp we had the pleasure of hosting the new Minister for Disabilities The Hon. John Ajaka. Muscular Dystrophy NSW was fortunate to have had such a good relationship with his predecessor, Minister Andrew Constance, and we were keen to meet with him early to raise his awareness of neuromuscular conditions and ensure this excellent relationship is continued. Minister Ajaka enjoyed watching the campers take part in Survivor Olympics and later addressed our Board, staff and supporters at a morning tea.

Over the past few months and following our *Client and Family Survey* we have been looking at what services we currently

provide, where there are gaps and where more support is needed. As a result we have implemented two new programs. The first is for those with a neuromuscular condition over the age of fifty. We launched this program in October with a lunch in Sydney where we were privileged to have our patron, Her Excellency, Marie Bashir join us. The second new program, Working Wheels, is an employment program for people with a neuromuscular condition where ADHC has generously funded us to carry out a two year pilot. For some time now you have been telling us about the challenges of finding employment and as a result we have developed what we hope will be a successful and ongoing employment program.

I am continually amazed at the generosity of our members and supporters. We value and appreciate your donations, your volunteering and the events you hold on our behalf which raises much needed funds. We couldn't provide the services we do without your ongoing support.

Pene Hodge
CEO, Muscular Dystrophy NSW



A warm welcome to Mel

Hey, I'm Melanie. I am currently in year 12 and have just started the HSC. Hopefully the next 12 months will go by quick so I can do some fun things! In my free time I like to watch movies and read books. I read and watch all sorts of things but my favourite genres are mystery and crime investigation. Sometimes I like watching horror films but I always end up regretting that I watched it!

"I have just recently been employed by Muscular Dystrophy NSW as their Web Administrator."

I am responsible for adding new events, news articles, flyers and brochures on to the website as well as keeping everything up to date. I have been working as a

volunteer for two years helping MDNSW update their website but it's great to now be actually employed.

"MDNSW is like a big family and being able to be a part of this wonderful family is a huge privilege for me."

The team has been incredibly welcoming from the very first day and I am honoured to be able to work with such amazing and passionate people! Every staff member at MDNSW puts in so much time and effort into creating amazing programs and opportunities for people who have a disability. I'm so glad I am able to contribute and help raise awareness of neuromuscular conditions through the website.



Apart from working with MDNSW, I am also undertaking my Gold Duke of Ed Award and I attended MDNSW's Camp Kula N' Gadu for the first time as part of my Residential Project. I was really excited to meet new people at camp and had a fantastic week of adventure and fun!

Melanie Tran
MDNSW Website Administrator



People for an Australian Muscular Dystrophy Day Saturday 30 November, 2013

*Be aware: wear a Red Ribbon
to show you care*

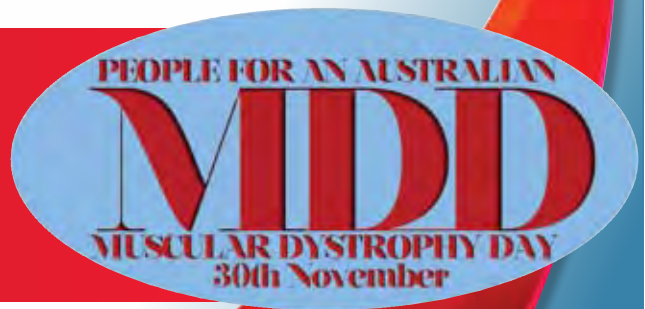


People for an Australian Muscular Dystrophy Day was a vision of Michael Baptiste who started a Facebook page in November 2011 hoping to have one day each year set aside dedicated to people with a neuromuscular condition.

Celebration Morning Tea

To celebrate this day you are invited to attend a morning tea at 10.30am on Thursday 28th November at the Muscular Dystrophy NSW offices, 5 Bay Drive, Meadowbank.

RSVP to Maralyn McCann (02) 9809 2111
or maralyn.mccann@mdnsw.org.au



Or host your own celebration morning tea at a location of your choice

Our MDNSW men play for Australia in Boccia Championships

Just three years ago I turned up for a local Boccia Come & Try Day hosted by Loretta Downie and MDNSW. Last week I captained Australia in an international championship featuring athletes from 9 countries. A lot can happen in three years! Of course a lot of things have changed. I train three or more hours a day, six days a week. I've travelled interstate and overseas to compete. I've taken part in numerous training camps in NSW and Queensland. I've met some major sports stars. I've presented medals and oh.... I've played for my country! None of these are things I'd ever dreamed of doing. But I've done them. If you enjoy playing just for the fun of it, go for it and enjoy yourself! If you do decide to go on to representative level though you'll need to be smart. It's not all about putting balls out on court. There's so much strategy involved. And patience. And concentration. And focus. You'll need these qualities when you play against the best players in the world like the Aussie Thunder did last week.

“ Nothing can prepare you for seeing yourself on a huge screen, or hearing the national anthem, or sitting out on a court on your own in a vast stadium with no one else but a referee and the world's number one boccia player next to you. ”



Tait Jenkins, 17yrs

Three of the twelve Australian team members have come through the MDNSW Boccia program: myself from the Central Coast, Dan Michel from Sydney and Tait Jenkins from Lismore. Jean-Paul LaFontaine from MDQLD



Captain of the Australian Boccia Team, Dean Nottle

makes it four players with MD in the Aussie squad! We're aiming to play in the World Cup in 2014, the World Championships in 2015, and ultimately the Paralympic Games in Rio in 2016!

Dean Nottle
MDNSW member

Working Wheels - do you want to be employed?

MDNSW is proud to announce that a recent proposal for funding submitted to ADHC has been approved and commenced as a pilot project in October 2013! Working Wheels is a self directed employment option for people living with a neuromuscular condition. This project is a world first initiative for people living with a neuromuscular condition aiming to ensure people have the skills necessary to move into the workforce, and support them and their employers through this transition.

With the phenomenal success of The Duke of Edinburgh's Award, we identified a gap in services available to assist many young people to obtain and maintain meaningful employment. Sadly many people living with a neuromuscular condition do not get the opportunity to experience employment due to poor accessibility, limited access to adaptive equipment, community attitudes and awareness, in addition to their limited physical ability that restricts their employment choices.

Working Wheels will address these issues and highlight that, given the opportunity, people with a neuromuscular condition can find jobs suited to their skills, abilities and interests and make significant contributions to their chosen workplace.

We will be working closely with businesses to provide a Business Education Program (BizEd) to ensure that employers are skilled and have an extensive knowledge of neuromuscular conditions. Most importantly, they will be aware of the progression and ongoing support needs for specific conditions. The pilot project will commence with 7 participants and upon completion will look at expanding to ensure we can provide this valuable support to as many of our members as possible.

If you are interested in participating in the Working Wheels project, either as an employee or employer, please contact Melissa on melissa.wentworth-perry@mdnsw.org.au or 9809 2111.

Training Project

The MDNSW Training Project is continuing its way throughout Sydney and the regional areas of NSW. We now have two new facilitators on board who bring with them a wealth of knowledge! Sandra Holland is the Clinical nurse at Sydney Children's Hospital Randwick, Neuromuscular Clinic. She has been travelling throughout the regions sharing her expertise in supporting people with a neuromuscular condition. Carolyn Campbell-McLean is a Living Life My Way Champion and Disability Training & Communication Consultant. Along with her trusty assistant, Anthony Bucco, she has been delivering the training to people in the Sydney Metro Area.



Anthony and Carolyn with some of their training participants

The feedback we are receiving from those attending the training sessions has been exceptional! Words such as amazing, informative, valuable and powerful are frequently appearing on the evaluations. It is great to see that so many service providers, OT's, physios, case managers, support workers and others are attending these training sessions and gaining an in-depth understanding of neuromuscular conditions. Our online e-learning tool is also continuing to reach a diverse range of people, with statistics showing hundreds of people have already accessed it online! The training project will continue to travel throughout NSW for the remainder of the year and hopefully into next year, with exclusive sessions being offered to those who are interested.

MDNSW receives another Award!

Congratulations to MDNSW for receiving yet another Award! This time it was for the 2013 NSW Carers Award which acknowledges and celebrates the outstanding contribution carers make for people in NSW. MDNSW was particularly acknowledged for

the quality of our Retreats and the state-wide regional programs we conduct, and we are immensely proud of all the staff who have worked so hard to ensure that carers are acknowledged and supported as much as possible here at MDNSW!

Registries: connecting patients and trials

Registries, or databases of patient information, are often sold as tools for research – but the important question is what can they offer patients? Neuromuscular disorder registries in Australia and New Zealand are two examples of initiatives driven by patient groups, which provide opportunities for people living with neuromuscular disorders to be involved in international clinical trials.

Registries contain clinical information on patients and are an important source of data for researchers to advance the understanding of a given disease, and enable monitoring and follow-up over time. The Australian Neuromuscular Disorders (ANMD) and the New Zealand Neuromuscular Disorders (NZNMD) Registries, established in 2010, have been strongly driven by patient support and advocacy groups including the Duchenne Foundation, the Muscular Dystrophy Foundation and Muscular Dystrophy Australia. These national overarching registries bring together information from disease specific registries for Duchenne muscular dystrophy, spinal muscular atrophy and myotonic dystrophy conditions.

The ANMD and NZNMD Registries contain patient's clinical and genetic data and assist in the recruitment process for Australian and New Zealand patients into international multicentre clinical trials. The registries are linked into the TREAT-NMD (Translational Research in Europe for the Assessment and Treatment of Neuromuscular Disease) global network of national registries. TREAT-NMD enables researchers, clinicians and clinical trial sponsors to identify individuals suitable for studies across multiple countries. This means that the Australian and New Zealand registries can connect

Australians and New Zealanders with neuromuscular diseases with international clinical trials, which has not been possible before now. As such, these registries help connect patients with treatments that are under development, and not yet available anywhere else.

The power of this approach was seen when in early 2013, TREAT-NMD coordinated the collection of patient data across the globe in response to an industry-led enquiry. The Australian and New Zealand Registries collated de-identified data and provided this to the TREAT-NMD database. Within 24 days, TREAT-NMD had collated information on patients and clinical centres around the globe specific to this enquiry.

The Australian and New Zealand Registries are open to anyone affected by these rare neuromuscular diseases, and registration is entirely voluntary. The clinical and genetic information about each patient is entered locally by patients' primary physician, in association with the genetic testing laboratories in each state. Information provided to TREAT-NMD is de-identified so that personal information such as name and date of birth are removed, and only clinical and genetic data are made available to consider whether a patient can be offered a place in a trial. Any patients eligible for a clinical trial are informed about new trials and studies through their primary care physician.

If you or someone you care for is interested in being involved in the Australian Registry, or wants to find out more, please talk to your doctor and visit www.nmdregistry.com.au

Letters to the Editor

Cory Tierney's story in the last issue of Talking Point was a great story and really showcased that committing to a regular therapy program can have positive results. We are really pleased for Cory and his family. It shows that there are always lots of different ideas and new treatment options.

Whenever taking on a new treatment the family should be asking about a few key things, including experience with kids, experience with the condition, goal setting and expectations of the treatment. It is also important to have discussions about the relative safety of any new treatment with treating medical teams. It is possible that two treatments are unknowingly

counteracting each other, or that overdoing a particular technique could be harmful.

All treating teams are supportive of families' individual treatment decisions as they understand each family's circumstances and wishes for their child are different. Open discussion with all involved will result in the best outcome for the child, and the team is always happy to be contacted with any questions or for information.

The Neurogenetics Clinic

The Children's Hospital at Westmead

Letters to the editor are encouraged. All submissions are to be sent to MDNSW CEO Pene Hodge at pene.hodge@mdnsw.org.au



Zebrafish get hooked on DMD Research

Duchenne Muscular Dystrophy (DMD) as well as Becker Muscular Dystrophy (BMD) are provoked by mutations in a gene named dystrophin. Due to the large size of dystrophin, the affiliated muscle diseases are one of the most common genetic diseases. No resolutive cure exists. Therefore, in our research department at the Australian Regenerative Medicine Institute, we are focusing on the generation and exploitation of animal models to get further insights in the onset and molecular network that cause the symptoms seen in DMD and BMD.

In research, we rely on animal models that replicate human diseases. While animal models can never replicate 100% of the aspects of human diseases, they allow researchers to conduct experiments that otherwise can not be performed. In recent years a particular fresh water fish called zebrafish has become an excellent model system to replicate human diseases. Even though from a broad perspective fish do not seem to be closely related to humans, zebrafish cells and tissues have not significantly changed in the time course of evolution. In addition, zebrafish have many advantages as a model system. They are about 3-4 cm in size and can be reared very cost-efficiently in small freshwater tanks (as seen in our aquarium). After 2 to 3 months, zebrafish start breeding and produce 100-200 eggs every week with embryos that develop very rapidly. Within 24 hours the brain, eyes and muscle are in place and the heart starts pulsating after 26 hours. Importantly, the transparent embryos develop in eggs, which enables researchers to readily observe them under the microscope over long periods of time or manipulate them experimentally.

At the Australian Regenerative Medicine Institute we have established a zebrafish model for DMD by mutating dystrophin in the fish, the same gene that causes DMD and

BMD in patients. As our recent research has demonstrated, this particular dystrophin-deficient fish model replicates the human condition very accurately. For the first time, this allowed us to film the detachment of a dystrophin-deficient muscle fibre in real time in a living animal (see movie stills below). Encouraged by this positive outcome, we are looking at the dystrophin-deficient model to gain new insights into the molecular and cell biological consequences of specific dystrophin mutations and evaluate therapeutic strategies in life animals. We have conducted experiments that have

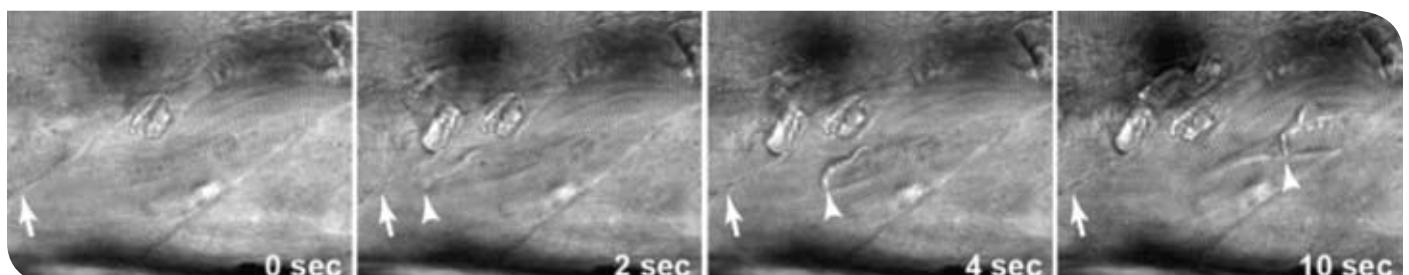
rescued the fish model by a recent approach called exon skipping. This technique manipulates the process, by which the information encoded by the gene dystrophin is translated into a molecular machinery. The outcome of this experiment not only allowed us to quantify the amount of dystrophin that in patients is likely to improve symptoms, but also demonstrated value of the dystrophin-deficient zebrafish model for the evaluation of therapeutic strategies. We are now in the process of exploiting

our dystrophin-deficient zebrafish model in order to find novel drugs. In a so-called small molecule screen, we use large libraries of functionally unknown chemicals that we test for any effects on the dystrophin-deficient fish. Once a chemical has been identified to improve the zebrafish dystrophic muscle, further research can concentrate on this particular chemical to analyse its use for potential drug development. While a cure for DMD might still not be accessible, many researchers all over the world – including us – are heavily investigating options and new therapeutic pathways to one day be able to provide patients better therapeutic pathways.

Joe Berger Ph.D., M.Sc. in Chemistry, M.Sc. in Biology



Zebrafish that carry a mutation in the dystrophin gene are swimming in freshwater tanks.



Consecutive images capture a detaching dystrophic muscle fibre in a living animal in real time. The arrow marks to the detaching point and the arrowhead points at the end of the detaching muscle fibre.



Sydney Metro Support Group meets again!

On the 31st of August 2013, I attended the Sydney Metro Support Group at Carolyn Campbell-McLean's home in Rydalmere. It was a nice outing to share with others over oriental dishes that each of us brought to the event. However, I may have missed that memo about the oriental theme as I took a cheesecake from the Cheese Cake shop that turned out to be a blast along with all the other fabulous food. The evening was a nice intimate setting where our small group shared our experiences and gained additional knowledge about access and equipment etc. Carolyn was a great host and made members of the Sydney Metro Support Group and their families most welcome.

"For me, the value of this regular event is the support network and contact with others with similar challenges to my own."



Carolyn expressed how beneficial it has been to share our stories and experiences over a meal at each other's home, where each person feels comfortable and relaxed with other members of the Group.

Hayley Bellamy, MDNSW member



An Over 50's Luncheon with Her Excellency



It was an honour to have Her Excellency, Marie Bashir attend our Luncheon



Ellie chatting to some of the 26 guests who attended the Over 50's Luncheon at Burnt Orange

My husband David and I had the absolute pleasure of attending MDNSW's inaugural Over 50's Luncheon on 22 October at the breathtaking Burnt Orange Cafe in beautiful Mosman. The building that houses Burnt Orange was built as the golf clubhouse for Mosman Golf Club in the 1920's. The Cafe is housed in a beautiful sandstone mansion overlooking Sydney harbour and located on a wraparound verandah with stunning views of Sydney Harbour, Mosman Bay and Manly. The Luncheon was attended by MDNSW Patron Her Excellency, Marie Bashir, Governor of NSW. It was a real honour to meet Her Excellency and we found her to be a very warm and humble person. We spent a lovely afternoon enjoying delicious food and the wonderful company of our fellow MDNSW members that we knew from before, and others that we had the pleasure of meeting for the first time. What a wonderful way to celebrate being over 50! Thank you to all involved in organising this luncheon. It made us feel very special and was very much appreciated!

Ellie Robertson, MDNSW member



Rick Arnheim, MDNSW Board member and Bruce Ellison catch up over a cuppa