

MUSCULAR DYSTROPHY
NEW SOUTH WALES

camp 2011

Camp comments



Parents of campers

"After a very nervous start and with some trepidation, I sent my daughter Bianca on her first camp experience. Hoping that she would make new friends and cope without me nearby, I was relieved to hear the happiness in her voice each time I spoke to her by phone. That week Bianca had lots of new experiences and got the courage to try new things that she would never have done before. This was possible because she was supported by caring, professional staff who included her, welcomed her, and made her feel she belonged – acceptance she hasn't always received from others in day-to-day life. It was a fantastic experience for Bianca and we are both truly grateful for it. A few days to rest and relax for myself was also something I had never done before, and very much appreciated. I discovered you don't really realise how much you need a rest until you have one! A big thank you to everyone involved, and God bless you all. You are all very special people!" **Denise**

"This turned out to be an exceptional experience for us as a family. It was the first time our 4 year old triplets have been with that many other people in a wheelchair, just like them. This camp is a rare, one in a million opportunity that we have enjoyed nowhere else. It was a fun experience which normalised things for our kids. My husband and I also benefited greatly from the camp. We spent the week being able to be spontaneous and carefree. We went out to dinner for the first time in ages; we saw a movie or two; we were able to live like "normal" couples – at least for a week! I had just gotten used to it and it was over!!" **Jemimah**

"According to Philip, the MD camp is one of the highlights of the year, something he looks so much forward to and something that just can't be missed. So imagine my horror when I found out in April that camp was already fully booked! Loretta promised to put Philip's name on the waiting list and luckily in May Philip had secured a place due to a cancellation. It's just so fantastic to know Philip is in good hands, having the best of times with friends, while being away from his family, and at the same time we can enjoy some respite and uninterrupted sleep for a week. It's always a joy to pick him up at the end of the week and hear all about his adventures and see the huge smile on his face. So we would like to express our huge gratitude to Loretta and all the fantastic staff and carers who make it all possible and rest assured that we will not wait even one second before we RSVP for next year's camp!" **Peter and Lilian**

Camp parent carer

"My son realises how lucky he is compared to some of the other campers, and how everyone is different and has different needs – but that everyone likes to smile and have fun. As a parent carer on camp, it's a privilege to be there and experience the joy and fun the campers receive, and see their confidence soar! It is a vital component of their life experience." **Vicki**

UWS camp carers

"Camp was one of the best learning experiences of my life. The children got to participate in amazing activities that they would not normally do and the smiles of pure delight was beautiful to see. My experience as an occupational therapist was invaluable to me. A text book could never teach me the skills I learnt about unconditional love, determination, will power and strength. The camp gave me the best hands on approach that anyone could experience and I recommend the camp to everyone!"
Rebekka Myles

The Camp was an exciting and challenging new experience which has allowed me to grow and develop as a future healthcare worker. Each day brought a different variety of new challenges on camp ranging from getting to know your camper, to encouraging your camper to participate in the days activities and the challenge of caring for someone else's child who has a disability and making sure they are safe and enjoying themselves. Throughout these experiences, the level of support and assistance throughout the camp by Muscular Dystrophy staff and by other volunteer workers was amazing.
Rebecca Meani

Campers' comments

"Camp was fun. It was a great experience because there was so much to do. Everyone was so nice and they all encouraged me to do things I never thought I would do."
Bianca, 17yrs old



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What a year it's been for events in 2011!

Our kids attended some Boccia Camps and qualified for the State Titles; they went to the Royal Easter Show, experienced the wonders of Dubbo Zoo, got to go to camp for 5 days without mum and dad, and showed the world what they are capable of doing in the Duke of Edinburgh's Award pilot program.

Our adult members attended our first Gala Ball at Doltone House, went on a weekend Retreat at Star City to see Dr Zhivago, met new friends at our New Members' Evening, flew to Melbourne for the weekend for some fine dining, saw the Blues get up in the State of Origin, learned about the latest research at the Medical Seminar, and caught up with the girls at the Ladies' Luncheon at the Waterview Convention Centre.

Next year we'd like to develop the sport of Boccia and the skill of Radio Controlled Car Racing throughout NSW. Our Camp and Retreat Program will be doubled and our Duke of Ed program will take on even more

recruits! You'll also see the return of some of our more popular events attended this year, with maybe a couple of new ones thrown in.

Remember that YOUR attendance determines which events we run and which events we don't.

I'd like to take this opportunity to say what an absolute pleasure it has been to implement this past year's events and programs for you all. I have met a heap of warm and wonderful people along the way and have been enormously proud of the many achievements I've been fortunate enough to witness. I'd like to wish each and every one of you a safe, healthy and happy Christmas, with lots of memorable experiences and special moments to come in the New Year.

Loretta Downie
Event Manager



Upcoming Christmas Celebrations

Sat 3 Dec Hunter Christmas Party
Light up Christmas in the Gardens

Fri 9 Dec South Coast Christmas Party
Christmas Cheers at the Winery

Sun 11 Dec Sydney Metro Christmas Party
Family fun and games hosted by the staff at CBA Direct Banking

Fri 16 Dec Far North Coast Christmas Party
A laughing lunch at Ballina RSL with a private Comedy Show

**REGISTER YOUR INTEREST
NOW FOR OUR CAMP
PROGRAM IN 2012!
DON'T MISS OUT NEXT YEAR!!**

Wed 18 April – Sun 22 April, 2012
One Mile Beach, Port Stephens

Mon 24 Sept – Fri 28 Sept, 2012
Sydney Academy of Sport, Narrabeen



Regional Services Update

Newcastle (Upper Hunter Region) Far North Coast (Ballina Region)

Have you heard of Leapfrog Ability?

LeapFrog Ability provides rapid response, flexible support services to people and families living with a disability in the Cessnock, Newcastle, Upper Hunter and Maitland areas.

LeapFrog Ability is a not-for-profit, non-government organisation guided by highly skilled and respected Board Members who donate their time, assets and efforts to benefit the community.

Leapfrog ability provides two great services clients living or caring for someone with a neuromuscular disease can enquire about. The Flexible Support Options and Community Living Program provide both "one off" or short term services such as

- Transport
- Equipment/Aids
- Household Items, White goods
- Independent Living Training
- Therapy
- Personal Care
- Respite Care
- Counselling, Behaviour Intervention
- Vacation Care
- Community Access
- Removalist
- Training services.



If you are interested in finding out more information about these programs, please call Leapfrog Ability on 4979 7777 or Webpage: www.leapfrog.org.au

New! Mother's Support Group

Far North Coast (Ballina Region)

This mothers' support group established in February this year invites mothers living in the Lismore/ Ballina surrounding area to join them for laughs, good conversations and morning tea!

The group, currently run by local parents caring for children with a neuromuscular disease, is eager to attract other mums to join this friendly, relaxed and informal network. Meetings are held once a month. Occasionally guest speakers from local networks are invited to discuss particular topics or discuss services available in the region.

If you are interested in finding out more about this support group please contact Kerrie Rogers on 0407 662 393 or email brogers2@bigpond.com

Remembering Dave

The 21st August 2011 marked the 10th Anniversary of our son David's passing to Muscular Dystrophy.

As it was a milestone anniversary, the youngest of our remaining three children, Louise, suggested to my husband and I that we try to do something special. She discussed it with her brother, Rod and sister, Melinda and their families and everybody came on board to help organise the day. Between all of us we decided on a family and friends fundraiser, with people donating whatever they wished. Those present took part in a walk, followed by a BBQ, at which they could purchase steak and sausage sandwiches, soft drinks and lots of delicious sweeties baked by our own family, plus a few special extended family and friends. We gave the day the title of "REMEMBERING DAVE" and invited people who had been close to David.

The 21st fell on a Sunday, which was great, and it turned out to be a glorious day weatherwise. We had decided



to hold the event at Stewart Park, right on Wollongong's beautiful beaches. The route for the walk was a very picturesque one and it took in WIN Stadium, the home of David's beloved Steelers and Dragons NRL team.

The family were there very early to grab the biggest hut and decorate it with a huge sign and lots of red and white decorations. We were delighted when 125 people turned up, some wearing Steelers and Dragons jumpers and some donning red MD tee shirts to wear on the walk. A few friends stayed at the venue to start the BBQ, so that by the time we returned from the walk we could start selling.

It was truly a wonderful day with everyone gathering together to remember David and support MD research. On the day we did very well financially, we also received sponsorship money from my nephew, his wife and some dear friends of ours who took part in another event to raise money. Altogether we raised \$3,000, which we were very impressed with, but the main thing was everyone remembering with love and laughter our beautiful boy. Our David.





regional events 2011

For many of you, you're probably wondering how quickly this year has flown by - well I'm wondering the same thing! This year's been a busy 12 months, with over 12 regional visits, many home visits undertaken and lots of fabulous metro events attended. I'm particularly thinking of how fabulous the Muscular Dystrophy NSW Ball was at Sydney's Doltone House earlier this year! It's been a great year. I hope you think so too!

With the New Year around the corner, you may be wondering what Carers' Regional events are being planned next year. Some of you may have suggestions for a guest speaker, social event or a topic you would like information about. If so I would like to hear from you with suggestions to better improve the current Care for Carers

Program. You can email or call me with your suggestions. I will be working on the 2012 calendar over the next few months. If you are keen on a particular idea or have any thoughts about regional events. I would love to hear from you.

As this year ends and a New Year begins, I would like to wish you all a wonderful 2012! May you enjoy the Christmas festivities planned with family or enjoy the wonderful holiday planned over the January holidays.

Georgina Christofis
Carers Program Coordinator

Young Carers' Spot!



Liam Cox is no ordinary seven year old. Nominated for the NSW Carers Award, Liam is a very special kid as described by his school teacher. Liam is great support to both his parents who are both in wheelchairs. His mother, Kelly, described Liam as "being a huge help". She said "He makes our lives easier that he doesn't even realise it." Kelly is so proud of her son, she is certain he will turn into a very sensitive and compassionate man when he's older.

"Although Liam didn't win the NSW Carers Award this year, he's certainly won over our hearts - well done Liam!"

Farewell Georgina!

Farewell to our Carers' Program Coordinator Georgina Christofis.

Thank you Georgina for all your years of service to people with Muscular Dystrophy, their carers and families. We wish you all the very best for the future.



Development of Treatments for Inherited Neuromuscular Disorders

We now know the genetic cause for many inherited Neuromuscular disorders, so why is it taking so long to find effective treatments or a cure? This is a frequent question at Neuromuscular clinics and remains a frustration for many people with Neuromuscular disorders.

We have accurate genetic diagnostic tests for most of the common inherited Neuromuscular disorders and these can be performed in Australian laboratories. However, testing for many of the less common disorders often has to be performed at overseas laboratories. Unfortunately there is no Medicare subsidy for many genetic tests and these are currently expensive with some testing costing up to \$8000. This is a significant problem as hospital departments are not funded to obtain genetic testing. The federal government has announced a consultation regarding genetic testing so there is a chance that this important problem will be addressed in the next 2-3 years. The other development that will lead to better and cheaper testing is Next Generation gene sequencing. This allows sequencing of the whole exome (most of the genetic material used for coding proteins) for about \$2000. This process produces such large amounts of information that it is still difficult to process all the information obtained but progress will be rapid. It also generates a lot of extra information and one of the challenges of Next Gen sequencing is deciding how to deal with this extra genetic information, or whether to ignore it.

Once the genetic defect is known, the next task is to work out the affect on the function of nerve and/or muscle. The biochemical pathways that underlie the functioning of cells and tissues are often very complex and it may take many years for scientists to be certain how and why a genetic abnormality results in a clinical disorder. Once this

is determined it is then possible to consider how to correct the defect.

One of the first steps is to reproduce the defect in an experimental model, which is usually done by introducing the abnormal human gene. The most frequently used models involve tissue culture of human muscle, rodents or zebra fish, however none of these are ideal. Tissue culture is problematic as the cells grow on a culture plate fed with an artificial medium, and are separated from other tissues. It is often crucial that they are associated with blood vessels and nerve tissue interaction to demonstrate the disease process. Both zebra fish and rodents may be useful models for human disease but can produce misleading results. The first step is to create something that looks and acts like the human disorder in the test model – this is known as producing a phenotype. Animal models with phenotypes (including rodents and dogs) for Duhenne and Becker dystrophy have been around for Duchenne and Becker Dystrophy for about 20 years and for about 10 years for Myotonic Dystrophy. While we have potential rodent models for FSH dystrophy we do not yet have one that demonstrates a definite phenotype.

Once we have an animal model, we can begin the process of finding a treatment, either drug-based or genetic, that will result in "cure" or improvement of the phenotype in the disease model.

Unfortunately, a treatment that is effective in a tissue culture or animal model may not be either safe or effective in humans. Rodents and zebrafish are short lived and tissue culture live for a few days only, so the life span of the disease model is short. This means that they may die long before developing evidence for the toxicity of a treatment. While muscular



*Prof. Alastair Corbett
Muscular Dystrophy NSW
Medical Director*

dystrophies may shorten life span, they are not rapidly fatal disorders and patients even with severe muscular dystrophies live for many years and lead enjoyable and productive lives. Therefore we are not justified in using treatments that are dangerous or may result in serious side effects, or be potentially life threatening. This is in contrast to some treatments for cancer. Many cancers are likely to prove fatal within months if not treated, so we can have a lower threshold with regard to the safety of medication. Medication or gene therapies for Neuromuscular disorders must be considered both safe and effective before introduced for general use. Ensuring effectiveness and safety is a long and involved process. Genetic treatments – exon skipping and stop codon read through - are now being tested in Duchenne muscular dystrophy. We do not yet know the effectiveness or possible long term toxicity and we await the outcome of these blinded and controlled trials.

At the present time, we can cure Myotonic dystrophy in the mouse model and work is proceeding on genetically curing it in human tissue culture using a short sequence of DNA called an Antisense Oligonucleotide. This gets into the cell and unblocks the

cell machinery that was malfunctioning due to accumulation of the expanded CTG repeats present in Myotonic dystrophy. If it continues to prove safe and effective it may need to be looked at in other Myotonic dystrophy disease models closer to humans before human trials can start. FSHD treatment investigation is not as advanced.

I am frequently asked why we don't use stem cells. There are many different types of stem cells and our own muscles contain stem cells which allow them to recover after damage. In inherited Neuromuscular disorders, our own stem cells have already failed. We can either use stem cells from another person (a tissue graft that might be rejected) or genetically modify the patient's own stem cells. We have 2

copies of each gene, a copy from each parent is inherited. There are 2 forms of inheritance - dominant and recessive. Recessive disorders occur when both copies (alleles) of the gene are abnormal - the gene product (protein) cannot be made and results in a loss of function. Autosomal dominant disorders usually occur because one allele makes an abnormal toxic product that causes damage to the cell resulting in a toxic gain of function. Stem cells could have a role in recessive disorders to replace the protein or function that is lost. They have been trialed in individual muscles in limb girdle dystrophies with some benefit. There is currently no way to administer muscle stem cells other than to inject them directly into muscle. Until some form of general

delivery by intravenous injection or similar becomes available, they are unlikely to prove useful. In dominant disorders, toxicity is problematic, due to an abnormal product from the defective allele. Adding stem cells is unlikely to do anything to counter this. The appropriate treatment is one which would suppress or counteract the abnormal gene or its toxic product.

We are entering an exciting time for patients and scientists with the very real prospect of effective treatment or cure. We should not however forget that conventional symptomatic treatment is also improving as we understand these disorders better, and that there is plenty to be done at the present time. We should not just wait for a cure.



On Wednesday 9 November, our Dukies all came together for a special lunch at the Novotel at Sydney Olympic Park following an invitation from the National Office of the Duke of Ed (DoE). The main aim of the lunch was for the DoE Ambassadors, Board and staff to meet each of our truly amazing young people who are doing their Award, and to also gain a better understanding of each person's individual journey. It was also a chance for Muscular Dystrophy NSW and the National Office of the Duke of Edinburgh's Award to acknowledge/celebrate the new partnership we have formed - ensuring young people with neuromuscular conditions have access to the Award program.

What a wonderful afternoon we all had! Sean started proceedings with an Acknowledgement of Country, and Hayley delivered yet another moving speech, outlining what her Adventurous Journey to the Pacific Islands taught her about how she wanted to live her life. There was a strong sense of Team as all the Dukies shared their journey achievements so far with the DoE guests, and also opened up about any challenges they had been having along the way. All in attendance were extremely impressed with our Team's drive and obvious enthusiasm for this pilot program, and Pene and I couldn't have been prouder of how each one of our Dukies rose to the occasion.

This is a strong team and we're really starting to make a huge



DoE Ambassadors and Board representatives with Members of Muscular Dystrophy NSW's DoE Team in their special edition jackets

difference together, which we hope will eventually pave the way for many more young people with a neuromuscular condition in the future.

Loretta Downie,
Event Manager



Harleys 4MD

On Sunday 16th October we held our new Harley event for the first time and it was an awesome day for all involved! It was a beautiful day and the sun was shining. We got to the Hogs Breath Café in Parramatta to the most amazing sight of all the bikes lined up, shining in the sun. Everyone grabbed a rider (or the rider grabbed them!) and we set off towards Windsor. The views along the way were stunning and when we stopped at the lovely Macquarie Park we were treated to morning tea – complete with great cakes!

We then had a great ride back to Parramatta where we ended our Harley ride and had lunch in Hog Breath Café. Everyone got to chat to the HOG's riders while we feasted on steak sandwiches and burgers. Hogs Breath Café was great as it really fitted the feel of the day and we sat in their outdoor area which was perfect in the hot and sunny weather. One of our supporters who came on the day was kind enough to write about her experience –

'Pulling in to the car park at the Hogs Breath café, I spotted them. Hard to miss - a big group of bikers, with leather jackets and waistcoats, heavily adorned with badges; black sunglasses and bandanas! I chose my bike - a huge, black, Harley with the iconic badge proudly standing out, shiny skeleton wing mirrors, leather seats and an exhaust which reflected the wonderful rays of the Sydney sunshine. I was introduced to my rider and he helped into my helmet, he gently clicked the safety strap into place and explained the signs for if I wanted to stop or slow down. He hopped onto the bike, and held out his hand, a gentleman helping me on to the back. I jumped on to the seat and he gunned the engine, signaling for the others to do the same. In unison, the 40 or so other Harley Davidson bikes started to move off towards the open road. Gracefully, the pack moved as one, the formation was quite the sight for the other vehicles and people walking by! After about 45 minutes we stopped for refreshments – cold bottles of water and a selection of cakes and

I got chatting to the others in the group. After a little rest we all hopped back on and headed back down the highway under the smiling stares of the people around us in their cars, heading back to where we started and a huge yummy lunch of burgers and chips. The feeling of the wind on your face, the sunshine on your back and the open road ahead of you is an experience I will never forget. They say you get the bug for riding a motorbike, well, until that day, I never understood it. I do now... a once in a lifetime experience...So, I wonder what colour would go best with my handbag?

The fact you can have that experience and at the same time support this wonderful charity, is almost too good to be true. And, it's a once in a lifetime experience which I urge you all to sign up to today.'
Rocky Brindley, 2011

'Loved the whole experience and like most I talked to can't wait for the next ride.' Donna Brown, 2011

We just want to say a huge thank you to everyone who supported the day. Thank you to Todd Le-Grand for organising the event and all the HOG's riders for giving up their time and coming to give everyone an exciting ride! Thank you to Hogs Breath Café for putting on an awesome lunch. And last but definitely not least, a huge thank you to everyone who came on the day, we really appreciate your support!

If you are interested in joining in the fun in our Harley event next year then please contact our Fundraising Manager, Kags Garrard, via email at kags.garrard@mdnsw.org.au or call the office on 9809 2111 and ask for Kags.



Fundraising Manager - Introduction

G'day! My name is Kags Garrard and I have just started in the role of Fundraising Manager. After 5 years of working for a children's charity in London, I have moved over to Sydney – which I have really settled into and already think is the best city in the world! I am really excited about my new role and developing the fundraising here. The week before I officially started, I was fortunate enough to visit one of our camps out at Narrabeen. What an incredible way to start and such an inspiring experience for me! Everyone involved, especially the children and families, were just awesome and I really enjoyed myself! Not many people get to spend their first day at work riding on a Harley Davidson!

Over the upcoming months, I am really looking for new and creative ways to fundraise, so please get in touch if you would

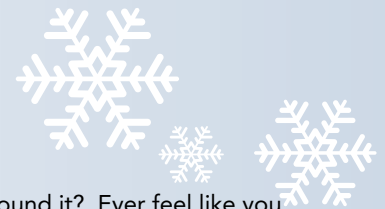
like to help out or you have any ideas – no matter how crazy they may seem! My door is always open for ideas and I will make sure that there will always be a smiling face and some coffee and biscuits waiting! I'm really looking forward to meeting you all in the future.

Merry Christmas!

Kags
Fundraising Manager



Do something wonderful this Christmas!



Do you love Christmas and all the activities that happen around it? Ever feel like you would like to do that little bit more for others or woke up with a great idea for a fundraising event or a way to make money? Ever sat in your office and thought about how you could get your staff involved and become more pro-active?

Well now is your perfect opportunity!

This Christmas, we are hoping that we can get lots of people involved with small fundraising projects and events all over New South Wales. Whether it involves carolling, Christmas parties, fetes or even donating that coin you find in your Christmas pudding! This is a great way to have fun and to help others in the process.

We can give you plenty of advice and support as well as send you lots of helpful fundraising material. If this is something you would like to do, then please contact our Fundraising Manager – Kags Garrard – on 9809 2111 or email kags.garrard@mdnsw.org.au

Volunteers Needed!

“ We are so lucky to have been supported by Spotless, who put our collection boxes in their businesses around Sydney International and Domestic airport. This fundraising and support is so fantastic and the partnership has made us close to \$40,000 per year, which I think you'll agree is absolutely incredible! We are looking for a volunteer or some volunteers to help us with this collection. This money is really vital to us so you would be doing such an important job if you get involved. If you would like to volunteer then please contact our Fundraising Manager - Kags Garrard on 02 9809 2111 or email kags.garrard@mdnsw.org.au ”

Thank you in advance!





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Yes, I would like to make a tax-deductible donation to help those with muscular dystrophy

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