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MDDA News

Newsletter of Metabolic Dietary Disorders Association

Issue 50
2012

THE AGE RUN MELBOURNE

Team MDDA @ RunMelbourne

On a beautiful Melbourne winters morning 12 Victorian MDDA members set off on out first MDDA Team Challenge: Run Melbourne 15th July 2012.

Ten of us chose the 5km event and enjoyed a lovely morning walking and chatting the course. Michael and Dean stepped up to the challenge and ran the 10km event. Well done guys!

This energetic group managed to fundraise over $2000.00. These funds will go to great use in supporting families and individuals who are affected by inborn errors of metabolism. We are looking forward to next July when we hope we can encourage more of you to get active and join in the fun with us!

Walking the path with an IEM!

Everyone's journey in life takes them different directions. The direction we often take is guided by those who have travelled before us. In life we look up to those who have experienced the highs and the lows. We try to learn from others mistakes and cherish the memories of those around us.

The path for those who have an Inborn Error of Metabolism isn't easy. Likewise for those who support these people it can sometimes be challenging. What is reassuring is that many people have walked the path before us. They have seen the obstacles and in so many ways are willing to share with us how to navigate the path.

In this edition of the newsletter we hear stories about joy. For some it was seeing peoples faces when their loaf of bread turned out perfectly at the **Cooking with Nutricia** workshop. For others it was the celebration of **Christmas in July @ QT Gold Coast.**

The member stories of **Reef Emmerson** and **Stella Contera** share with us how our newest members are doing. The moving story about **Tayla Richardson** (UCD) gives us an insight into her time with us.

The speech that Nicholas Mazzone gave at the **WA Pasta Lunch** educated those about what is involved for an eight year old to manage his diet.

We hear stories about raising awareness of IEMs in the community. In SA, Katie Graue was part of a **PKU Awareness Day** at her Kindy. We share the article **"Don't feed the boy protein; it can prove fatal"** that appeared in the Brisbane Times and was the 2nd highest read article online that day!

We get an update on **Dean Tulloch's North Face 100** Ultra-Marathon and hear what went on during the **Far North QLD Clinic Run**.

We finish the newsletter with an update on the **PKU Research at the Children's Hopsital at Westmead** headed by Professor John Christodoulou.

Richard Drewitt
MDDA Executive Committee Member

this issue

Donations over $2.00 are Tax Deductable

www.mdda.org.au

CORRECTION

In our last newsletter we had an article featuring the NSW Adult Clinic. The number listed was incorrect. You can reach the Westmead Adult Metabolic services on (02) 9845 9780

IEM GRANT

Just a reminder that to remain eligible for this grant you have individual responsibilities such as maintaining your diet as evidenced by regular blood tests and keeping clinic appointments. Failure to comply may mean your access to the grant could be removed. Any questions please contact the Department directly on (02) 6289 8980. Or contact us at the office.

AGM

With this edition of the Newsletter a flyer outlining the details for the AGM will be enclosed. If you have any questions please contact the office.

EXPRESSION OF INTEREST FOR PARENT PARTICIPANTS

RMIT University, School of Health Sciences, Honours Research – Meaning Making and Family Quality of Life

This project focuses on understanding families' experiences of quality of life and how they give meaning to their lives when experiencing stressful parenting. To participate in this study, all you need to do is complete an 'Expression of Interest for Parent Participants' and a very quick questionnaire. For full details, please contact Amanda Yee at s3359448@student.rmit.edu.au.

About the MDDA

The Metabolic Dietary Disorders Association is a national self-help group supporting people affected by genetic (inborn) errors of metabolism. Our aim is to offer families a comprehensive resource of information and support. We provide members and their family's forums to share experiences and information, to educate themselves and the general community about living with an inborn metabolic dietary disorder. The MDDA fosters co-operative relationships between members, healthcare professionals, government departments and other agencies to promote the health and well-being of members. MDDA objectives are pursued by the Committee of Management.

Cooking with Nutricia!

We love doing what we can to support the metabolic community and that is why we thoroughly enjoyed the Nutricia Brisbane Low Protein Cooking Workshop earlier this month!

Over 30 Queenslanders came to enjoy our cooking classes which were run by our low protein cooking expert Fiona Wedding. Some new low protein cooking stars came out and shined, successfully making breads, pasta bakes, muffins and all sorts of goodies. The advanced cooks got to try our new expert level recipes and succeeded with making low protein tarts, tortillas, patties and more.

The highlight of the day was seeing the surprise and joy on each person's face as their loaf of bread turned out perfectly! We also loved how many friendships formed over a bowl of risotto.

We love helping people bring variety to their diet as we know how important this is. You can make all sorts of delicious foods using our baking mix, breads, rice, pastas, biscuits and milks.

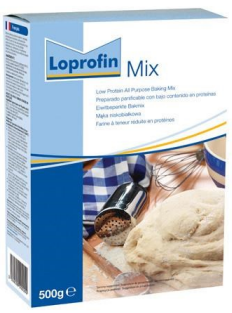
We know our range has become smaller recently and we understand your concerns. The good thing is that our remaining low protein products are very versatile and we want to show you just how much you can do with them.

Our website has over 80 different recipes and tips on how to use our breads and pastas for the best results. We'll also be coming out with a very exciting online surprise soon so stay tuned…

Until then, check out our website at www.nutrition4me.com.au and look under MyDiet for recipes and MyProducts for products! Also feel free to drop us a line under 'feedback' as we'd love to hear from you.

Happy cooking!

From the Nutricia Metabolics Team









Christmas in July @ QT Gold Coast

Putting the Fun in Fundraising

So there comes a time when you think you might have bitten off a little more than you can chew, your work life is hectic, your family commitments are piling up, your friends are desperate to see you because it feels like an age since you have graced them with your presence. What to do? I know, plan a fundraiser! "Yeah, Because you haven't got enough on" My Mum joked.

After trolling the social pages (which were uncharacteristically foreign to me these days) on the search to find a venue, I settled on the Bazaar Restaurant in the QT hotel on the Gold Coast. The idea behind this establishment is a modern take on a farmers markets. All the produce is fresh from the farmers that morning, or off the seafood trawlers, and they only serve seasonal produce to ensure the freshness of all the meals served so I knew all my foodie friends would be impressed. The decor is modern retro with a coastal surf theme giving a real laid back approach to what I thought would be a perfect environment for a fundraiser.

I had entertainment in the bag as my musician husband recently recorded with 2009 Australian Idol finalist "Casey Barnes" and Casey was so excited to do what he could to help. We are also close friends with some music industry talents and so we had a stella line up of entertainment for the day - including Santa Claus, who was on his winter holidays but helped out when he heard it was for the MDDA. I helped Mrs Claus wrap 80+ individual golf balls for the men coming and cosmetic gifts for all the women. Santa also gave the kiddies some gifts, which was the highlight of their day.

Then came the hard parts - I had to ask for help... I approached all the companies that I could think of to help donate items for raffles or prizes for give-away's. I went for big things and and I went for small things. I was prepared to be turned back every time. What I wasn't prepared for was the generosity that I received. It pays to ask for help. From kitchen equipment from the Good Guys to signed Gold Coast Suns Jerseys, I was overwhelmed with the response. There came a point where I had to group prizes together because we would have been raffling items off the whole day with no time for the entertainment. I have never felt a sense of community and belonging like I did in knowing that people were willing to help, I just had to ask.

I was struggling a little with selling tickets at the beginning, which I suppose is the way these things tend to go. We approached a local radio station and had an interview with the DJ talking about my sons condition (PKU) this was the perfect lead into announcing my Christmas In July themed fundraiser and from there we started to sell tickets.

The day came around before I knew it and started with the arrival of some faces I haven't seen for a long time, all my friends and family that I had promised to catch up with came along to help support me and the MDDA. My sons Kindergarten bought a whole table worth of tickets and made it their Christmas party. The raffle tickets were selling like hot cakes and the laughter and enjoyment was so great to see. From tables of ladies having a ladies luncheon to work Christmas parties - the venue was buzzing with excited energy on the prospect that Christmas was coming twice this year!

My husband taught himself how to make a movie on the program imovie and we showed a film to the guests of my story and a little information about PKU and what it all means. The film explained what it was like for me to receive the diagnosis of PKU for our little boy, the steps we took after that and the diet and medication that is required for him. The film was meant to be a informative documentary, but I think everyone in the room was touched by the raw realness of the film

(Continued on page 5)

Christmas in July @ QT Gold Coast cont.

(Continued from page 4)
and there wasn't a dry eye in the house. We passed a hat (santa lent us his) around the room and people were donating more and more to the cause.

During the film presentation I stood at the back of the room - as it was a little exposing for me to show this film. The waitress at a table who was collecting glasses at the time, stopped to watch and at the end of the film she came to me with tears in her eyes and told me I was an inspiration to her and she was touched by what I was doing to help my little boy and the association. She said she had never heard of PKU but that her partner worked with children and she thought she might take a brochure to him to see if he could spread the word some more for us.

Another of the guests on the day was a nurse at a local hospital. She came to me at the end of the day and expressed her feelings also about the heel prick test and how important it is. I told her how I remember thinking is this test really necessary - *"do you really need to take that much blood from my baby and how painful is this going to be to my little one, he is still so tender and new."* Quite obviously the view of a person with no knowledge of what this test if for. The nurse told me the lack of education about the importance of this test was mind boggling and commended me on spreading the word.

With the massive day coming to an end we packed up and de-briefed with a few of my close friends of a few much needed and well deserved cocktails. I was exhausted but on a high from the adrenaline of the day. My friends told me everywhere they looked people were raving about the food, service and how incredible the day was. The guests loved the raffle prizes and funnily enough all of the people who won the prizes where the people who truly deserved them,.

My mum rang me the day after the event to tell me how proud of me she was and that in years to come when Cayden is older he too will be proud. She said *"It was a wonderful day and no one felt like it was a fundraiser - more of a fun raiser. "*

Zoe Mitter

WA Pasta Lunch

On Sunday 20 May 2012 The Abruzzo Molise Sporting Club held a Pasta Lunch to raise funds for the MDDA. My 8 year old son Nicholas stood up at the beginning of the afternoon with his 2 cousins Lucas(7), Stella(1) who have PKU as well and spoke to approximately 200 people on the day. Nicholas also helped sell the homemade biscuits and raffle tickets. In total the Abruzzo Molise Sporting Club raised $1630.14.

Diana Mazzone

Hi My name is Nicholas and I am 8 years old and these are my cousins Lucas and Stella. We were diagnosed at birth with a metabolic disorder called Phenylketonuria most people know it as PKU. We can't eat a lot of things like meat, fish, chicken and eggs because our body can't break it down. Most of the food we eat needs to be weighed because we are on a strict low protein diet. After every meal we have to drink a special supplement which helps our body to grow strong. Our special diet is for the rest of our lives. It is very hard sometimes when we go to parties and out with friends because our parents need to bring everything for us.

Every month I need to have a blood test which is sent to Princess Margaret Hospital so the doctors and dieticians can work out if I need more supplements or if I can increase my food allowance. On behalf of the Metabolic Dietary Disorders Association Lucas, Stella and myself we would like to thank the Abruzzo Molise Sporting Club for hosting this lunch for us today to raise awareness about PKU.

Nicholas Mazzone

PKU Awareness Day (SA)

On Thursday 17th May 2012 my eldest daughter, Katie (PKU) Kindergarten supported Katie by raising awareness for PKU.

A newsletter was sent home to all families asking their children to wear something blue in recognition of PKU Awareness Month, and if possible make a gold coin donation with proceeds going to The MDDA. It was great to see almost all the children participate, and we raised $35 for The MDDA.

The director of the Kindy asked if I could talk to the children about PKU. The children loved telling me about what their favourite fruit and vegetables were to eat and how they helped their parents grow vegetables in the garden! Katie looked quiet proud hearing that her friends enjoyed eating a lot of her "free" foods. To keep it simple I read a book to the Kindy children, "Robin and I Explain PKU" by Jane Brummitt. They liked the pictures in the book as they were drawn by a young girl.

At the end of the day I gave each child a blue balloon to take home with a small information card attached explaining about PKU and The MDDA, so that the families of the children could learn little also.

Overall it was a great experience for both Katie and myself to see our community taking an interest and supporting PKU.

Cassie Graue

Don't feed the boy protein; it can prove fatal

At just over two years old, Cayden Mitter is like any other child his age. He loves planes and cars, can recognise 22 letters from the alphabet and is pretty sure the word "please" has magical properties.

To look at him, Cayden is normal in every way. Except protein could kill him.

The Mitters are one of about 700 Australian families living with a loved one who has a metabolic dietary disorder; essentially, inborn errors that stop the body from being able to process either fats, carbohydrates or protein or, in the unluckiest of cases, a combination.

In Queensland, it is estimated that just seven babies are born with a metabolic disorder each year. Phenylketonuria or PKU, the inability to process protein, is the most common metabolic disorder, "the one you want if you have to have one", Cayden's mother Zoe explained.

Each mouthful of food that passes through her son's lips must be monitored. Considered "high tolerance", Cayden can have 12 grams of protein a day, which means he can still indulge in the odd marshmallow or biscuit for a snack and is able to eat normal rice. But breads, cereals and anything derived from an animal is out for the rest of Cayden's life. Any more than 12 grams, or "points" as is the measurement the

Picture: Michelle Smith Source: Brisbane Times

Mitter family use, and Cayden will lose the ability to think clearly and be unable to concentrate. Prolonged exposure will lead to brain damage and eventual death. Those who have been diagnosed later in life rarely live beyond 50.

"We found out about it after we had the heel prick test done when Cayden was born," Mrs Mitter said. "It's not a mandatory test but we are so thankful we had it done. We were able to get Cayden started on his special diet and program as an infant. The damage that can be done to the brain is irreversible." Both Mrs Mitter and her husband Otto, carried the defective half of the gene that causes PKU.

The Gold Coast couple have been told they have a one in four chance of having a second child with the disorder. "We do know families who have two and even three children with PKU or another

(Continued on page 7)

Hash Brown

Ingredients
2 TBSP of oil
100 grams of shredded potatoes
100 grams of radish

Method
Oil a frying pan, and combine shredded potatoes and radish in the pan and cook for 5 minutes on low-medium heat. Stir and flip the hash brown mix until it is cooked through. Add salt & pepper for a crisp (optional) – I also like to add mild tomato salsa, ketchup, and/or mango peach salsa, but you can try adding any toppings that you like!

Hint
I think the more radish you add, the better. Using more radish and less shredded potatoes will result in a lower protein content and much healthier meal overall.

Don't feed the boy protein; it can prove fatal cont.

(Continued from page 6)

metabolic disorder," Mrs Mitter said. "I have no idea how they do it. It can be mind-boggling hard."

Cayden's "special food" is expensive and difficult to source, with most not meeting Australia's strict customs regulations. But Cayden must eat a certain amount of protein each day to aid his growth and development, even when sick. "Those are the most stressful times. When he doesn't want to eat. He has to eat. We don't have a choice," Mrs Mitter said. "But trying to convince a sick or grizzly toddler to eat is a challenge at the best of times. When their development counts on it, let's just say you have extra motivation to keep offering that spoon or bottle."

The Mitters have just cleared the first hurdle in Cayden's development, finding a kindergarten they can trust. "I was tearing my hair out, thinking about kindy and the food sharing which happens between kids," Mrs Mitter said. "We have been really lucky in that the one we found understands and have been amazing. They keep his food completely separate and have a no food sharing rule. But they include him in everything as well, so he doesn't feel left out. If there is cake, they tell me so I can make him a special cupcake and they put it on the same plate.

"But as he gets older, it gets harder. Next is school and then as a teenager … it is hard to understand unless you are living it. But it is not a case of because he can have a little bit of protein, it's OK. This is something he has to live with for the rest of his life."

The Mitters were directed to the Metabolic Dietary Disorders Association by their nutritionist, which Mrs Mitter called "an absolute lifesaver". The not-for-profit organisation provides support, recipes and advice for parents navigating their children through metabolic minefields. "No one knows what PKU or any of the other disorders are. Otto and I had no idea what it was when the doctor told us. Without the MDDA, we would be completely lost." In an effort to give back to the foundation and raise awareness of metabolic disorders within the general community "so waiters don't just think we are being overly picky and people understand why I won't let my son have a sausage at a barbecue", Mrs Mitter has organised a Christmas in July fund-raiser.

"It's not a food allergy. It's not us being hypersensitive. It is about our son's quality of life. If more people could become aware that these disorders exist and if we could raise funds for the organisation which guides so many of us through it at the same time, then I would feel that we have achieved something."

Amy Remeikis
Source: Brisbane Times 1/7/2012

http://www.brisbanetimes.com.au/national/dont-feed-the-boy-protein-it-can-prove-fatal-20120630-21a14.html

Bunnings BBQ @ Gold Coast

We all know that those with PKU or any of the other IEM's can't eat standard sausages however that didn't stop us from using the tried and tested technique to raise money. Recently at a Gold Coast Bunnings outlet I gathered a few friends and was successful in pulling off a BBQ which raised $976.

Zoe Mitter

Low Protein Crumpets

2 cups Loprofin Flour
1 sachet yeast
1 tsp salt
2 tsp baking powder
1 tsp sugar
1 Tbls oil (vegetable or olive)
2 cups warm water

Method

1. Mix together all dry ingredients.
2. Add oil and water and mix to a smooth batter.
3. Cover with cling wrap and allow to rest for approx. half hour in a warm place. The mixture should almost double in amount and look nice and bubbly.
4. Grease a large fryer pan and some egg rings to spoon the mixture into for a good round shape. Cook the crumpets over a moderate heat, as they work best when cooked relatively slowly. Use a toothpick to pop air bubbles as they rise.
5. Flip the crumpets over, only when mixture is almost dry on top, briefly in pan.
6. Freeze well. Toast in toaster when ready to eat.

Cass Graue

Member Stories

Reef Emmerson

Well Reef's has recently celebrated his 2nd Birthday! Its been a quick year full of challenges which are finally settling down!

Reef was diagnosed with Epilepsy not too long after his 1st Birthday, and fortunately this was picked up before any dramatic seizures, with the Metabolic Clinic actually noticing some strange symptoms at a Clinic appointment, which lead to investigations by the Neurology department; the hospital staff again proving how brilliant they are!

Reef's slow weight gain has continued to be an issue – a fussy eater who only eats tiny portions caused his body to break-down the stored protein in his muscles, releasing high PHE levels into his bloodstream. To control his PHE and put some weight on him, he was put onto Duocal with each supplement. This helped his levels and slightly improved his weight, but resulted in him only eating about 1 meal a day – if we were lucky!

On a positive note we eventually thought we would try him without the Duocal for a week or so, and his food intake dramatically increased, weight gain has been steady, and we are feeling much happier!

I am getting a little more confident with my cooking, but am definitely still always looking for some basic recipes for Reef, who still likes things kept simple.

We have successfully moved from heal-prick testing to finger-prick tests, and Reef has taken to the change well, as he was really starting to hate the heal-prick!

Developmentally Reef has done amazing this past year – he first walked at 18 months, and once he started we couldn't stop him! He is a very active little man who likes to be busy at all times, and his intelligence and vocabulary really is amazing. He is singing, counting to 10, naming his shapes and generally bossing everyone around far beyond his years!

We celebrated Reef's birthday at a playcentre with a huge group of family and friends. We barely saw Reef as there was too much fun to be had! As an alternative birthday cake I purchased Donut King Mini Iced Donuts which are 1gm of protein each and were of course popular to everyone!

Each year has had its challenges, though is getting easier every day, and we look forward to the challenges ahead!

Reef is now preparing for an exciting year of becoming a big brother which will prove to be interesting for him!

Wendi Emmerson

Stella Contera

On 14th May 2012 we celebrated Stella's 1st birthday. It is hard to believe a year has passed as it only seemed like yesterday that Stella entered our lives.

Stella has PKU like her brother Lucas (7). For her birthday, her dad David made a gorgeous pink fairy castle cake. The mini swiss rolls that sat on top of the cake had only 1gram of protein so Lucas, Stella and their cousin Nicholas (who also has PKU - yes you heard right there's 3 children with PKU in our family!) got to enjoy a piece of the cake too!

Stella is always complimented on how smiley and cheerful she is. She loves her PKU food and formula and is growing to be a very happy and healthy little girl.

Stella, like her brother Lucas, does very well with her PKU diet. Lucas conforms to his diet really well and loves to have his PKU Cooler. Even at the age of 7 he is already reading nutritional labels to work out whether he can or can't eat something.

Stella, although still so very young, shows a special connection with her brother Lucas and he is happy he gets to share his diet with his little sister. I have no doubt Lucas is a great role model for Stella as he shows a great spirit and attitude towards his PKU.

Maria Contera

Tayla Richardson
(19/06/2008 - 2/11/2011)

For me personally, the best aspect of being involved with the MDDA is speaking to other families and individuals affected by an inborn error of metabolism (IEM) and hearing about how they deal with their particular Disorder on a daily basis.

Generally speaking, despite the challenges we sometimes face, we seem to get on with it and do the best we can so that each person affected with an IEM can have the life they are capable of having and fulfill their potential. It's certainly not always easy and that's when our support network plays an important role.

Unfortunately when speaking to families on the rare occasion (very thankfully!) it's not an uplifting story but rather one that tugs at the heart strings and makes me realise just how precious life is and how important it is to be thankful for what we have. On those occasions, the story involves the death of a treasured child, leaving behind a devastated family trying to cope with the unthinkable.

Tayla Richardson aged 3, died on the 2nd of November 2011 whilst admitted at the Royal Children's Hospital (RCH) in Melbourne. Tayla was diagnosed with Urea Cycle Disorder only 7 months prior and her loving family had been on that roller-coaster ride we're familiar with of trying to understand what the diagnosis meant and do all they could to ensure her levels were maintained and her health was at its best.

According to Cath, Tayla coped well with her diagnosis, bravely bearing all those blood tests and not fussing when Cath had to change the tape on her G.N. tube on a daily basis. With a sweet personality, Tayla was full of love for everyone, and brightened each day. She loved spending time with her brother and sister and playing princesses. Like a lot of children on a

low-protein diet, Tayla loved her chips, and her mum, Cath knew just where to get the best ones.

Unfortunately we can't always control what goes on in the human body and Cath had to bring Tayla down from Piggoreet (near Ballarat, Victoria) to be admitted at RCH because she was too unwell to be treated at home. It wasn't the first time, and Cath wasn't unfamiliar with how it all worked. What should have been a successful outcome turned into a nightmare for

the Richardson family. It appears that as a result of an overdose of medication, Tayla never recovered and died in hospital four days later. It's not up to me to comment on the how's and the why's and to try and explain what happened during that admission, I'm sure these questions are still being asked by people today; rather I wanted to share the Richardson's story with you, at the request of Cath, so that we all have a chance to get to know who little Tayla was, and that we're aware of what can happen with the aim of preventing it from happening again.

Tayla is greatly missed by her older brother, Jeremy; sister, Molly; mum, Cath; and dad, Jason as well as her extended family and friends. I can't imagine how you begin to come to terms with the loss of a child, Tayla obviously greatly touched the lives of those she came into contact with in her very short life, and she is still greatly missed today.

Urea Cycle Disorder is a rare metabolic condition, unfortunately unlike PKU it is not detected via Newborn Screening. It is usually diagnosed when the child (or adult) becomes very unwell and presents at hospital. It is a genetic disorder caused by a mutation that results in a deficiency of one of the six enzymes in the urea cycle. These enzymes are responsible for removing ammonia from the blood stream.

Excessive ammonia in the blood can reach the brain and can cause irreversible brain damage, coma, and/or death. The onset and severity of urea cycle disorders is highly variable, depending on the specific mutation. The more severe the mutation, the more severe the urea cycle disorder. (http://www.nucdf.org)

Susi Hendricks
Vice President

Member Stories Wanted!

Do you have a story you wish to share? Forward us an email along with photos to **news@mdda.org.au**

Dean Tulloch's North Face 100
Race Report

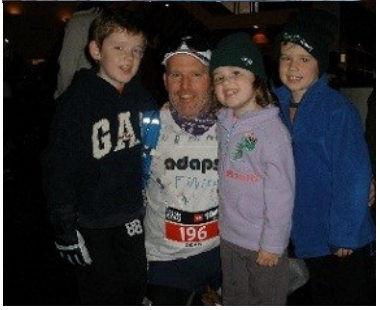
North Face 100 Complete!!

It was 19th May 2012 when MDDA Friend and Team MDDA Ambassador Dean Tulloch completed the North Face 100. The 100km race is a end to end ultra-marathon running through the beautiful Blue Mountains of NSW. Having completed the race in 18hrs and 40 mins Dean was greeted by eagerly awaiting wife and three kids. Dean crossed the finish line at 1.40am the following morning.

Dean has been an inspiration to us all, and his latest achievement has been enormous in generating enthusiasm and support for the MDDA extended community. The latest challenge followed on the back of his gruelling 250km Ultra-Marathon through the Gobi Desert in China last July 2011.

Dean has now raised over $40k and is continuing his journey to achieve his pledge of $50k (or more) to MDDA.

Thank You Dean for your support!



Conditions: Perfect. Could not have asked for better conditions. There wasn't any rain, and it wasn't cold - so perfect.

The Race: Technical. A lot of up and down - more like a hike with a little bit of running involved. My goal was to come in under 20 hours - and I came in at 18hrs 40mins - so very happy with that.

Start to Stage 1 (18km)

A lot of single trail (hard to pass people) and a lot of climbing up and down steps. A clear memory was the magnificent views that you get whilst running/jogging/hiking and climbing up the side of a mountain looking over the Jamieson Valley. Breathtaking.

Stage 1 to Stage 2 (20km)

The race opened up a little bit here. Started feeling it a bit more as we got closer to the 38km mark. At the 33km mark, a guy running behind me ran up to me and said "You were lucky then mate". I said "What are you talking about?". He said, "A 6ft Black Snake just slid out from one side of the track behind your legs to the other side". A snake !!!! Luckily I didn't see it or I would have freaked out.....(as if the race isn't hard enough). But we move on.

Stage 2 to Stage 3 (16km)

Pretty tough stage this one. Lots of rocks to jump over. Fell twice on this stage and felt the legs lock up both times and thought - no no no...please don't be injured....but luckily - no injury. Got to the end where my wife (Kerry) and kids (Flynn, Henry and Zara) were waiting. They were the support crew and was great to have them there.

Stage 3 to Stage 4 (11km)

After feeling totally exhausted at the end of Stage 3 (54km down) I actually took off pretty well. Had re-fuelled and felt alive. The first half of this stage was great - slight hill climb - but ever-so slight - and I ran pretty hard. The second half of this stage (5kmonly) was a different story however. Climb Climb Climb up the golden staircase. Absolute leg burner and thought it would never end - but it did ....and ran into Checkpoint 4 which was an indoor Aquatic Centre. Kerry and the kids were there as well, and again, chance to re-fuel.

Stage 4 to Stage 5 (24km)

Well this was always going to be a test. 65km down and this is a long stage. It was dark at this stage and a little cold - but pretty mild at the same time. The start of this stage is a lot of bush running and then tourist track running. You are guided by your headlamp and there reflective vests of other runners. Pretty cool to be running in the dark in the middle of nowhere. Started off strong but deteriorated as we started to go downhill. On this stage there is a lot of downhill running - and this really hurts the knees and in particular the ITB(Illiotibial Band) - You run through it for the first few kms but eventually the pain is pretty tough so my running turned it to a bit of a hobble and a shuffle - but eventually got to the bottom of the valley. Then, of course, we had to climb back up - and had between 8 -10km of uphill - which was just impossible to run - but much better on the knees- so didn't mind it. We had a mandatory bag check half way through this stage - which is good and bad - good that you get a quick rest and bad that you have to take your bag off search for stuff in the dark and get everything back together again - and after 80ks of going through the Blue Mountains all you want to do is keep going and finish. Eventually hobbled into checkpoint 5 (89km mark) in a lot of pain but equally excited that this thing was almost over.

Stage 5 to Finish (11km)

After a good break at the very cold Stage 5 checkpoint it was time to trudge off again. The beginning of this stage was on asphalt road -slight uphill incline. I tried to run - but was struggling. I found that I could run for say 20 metres and would then have to stop due to the knee pain. My style was very unorthodox to say the least - but who cares....just got to keep the legs going forward rather than not going at all. So we had a few km's of running on road at the top of the Blue Mountains - and - eventually we had to go back down - so we did. Down on tracks along the side of mountains and eventually back up again. Almost there? Well ...no. With 3 kms to go I am thinking where is this finish line? Maybe a straight road to the end would be good? No There was a 3km to go sign with an arrow that went straight down. Yes -time to head back down into the valley on large

STEPS. And walking on steps going downhill with bad knees is painful - and itw as. So this took me a fair bit of time to be honest. Couldn't go fast at all and was in a bit of a sideways motion. Then 2km down -still - down and across - this was a tough finish. Finally saw the 1kmto go sign and started climbing out (climbing up a lot easier than climbing down). Found the hotel and the track to the finish line and got my mojo back and started to skip/run like I've been riding a horse for 100 days straight and finished the race.....Kerry and the kids were there - bleary eyed at 1:40am in the morning...was fantastic and loved every minute of it. Then went upstairs and...crashed...

Dean Tulloch

Far North QLD Clinic Run

Well its that time of the year again, something I always look forward to each year. The Far North Qld Clinic Run May 2012 for Townsville and Cairns.

This year we had Dr David, super nurse Anita, Sarah and dietician Ming Liew as our fantastic metabolic team.

In Townsville on a busy Tuesday morning Miss Jaime-leigh Denison (18) (PKU) is the first to arrive, brother Luke (15)(PKU) will be picked up from school and come later. Jaime is studying Zoology at JCU, she loves getting the MDDA newsletters to read stories about other families and the new products on the back. She'd like to see more cooking demo's in Townsville. Luke is in yr 9, wants to be a chief when he leaves school, and his favourite food is lasagne. Casey Lucas (18)(PKU) is also studying at JCU doing primary education, her and her family went to Hawaii earlier this year, she said it was great and there were lots of varieties of foods to eat…Rhys & Bernadette both who are also studying at JCU have a beautiful 18mth old Mia, (pku) who's a little ball of energy, it sounds like JCU is the place to be this year.

A few other families Cale Guglimana and Rubie Allan,(galactosaemia) where in and out so quick we didn't have a chance to chat or get their photos. Troy Giles and mum Verona where there also, Troy (PKU) was telling me he already started to plan his 40th next Feb, can't wait to hear all about it at clinic 2013.

Lucas (PKU) and Allison were next, Lucas is 3 and loves vegie chips, low protein mac cheese & lasagne, and he has the most beautiful red hair. Lucas and Ryan where playing Angry Birds on Ryan's phone. Lucas is a typical little boy, he never sat still, I'd love just a quarter of his energy.

Kurtis' (PKU) mum Cassandra and Grandma travelled in from Charters Towers. Then in came the Bamford's Jo and son Angus 6 (PKU) he had a great day at school, he loves garlic bread, tomato base pasta & yellow pasta.

Alan, Madonna and Mitchell, came next, Mitchell had his 21st in February, how time flies, Mitchell was telling us about his new shoes, he also loves vegie chips. Ryan, Jaime and myself had a lovely chat with Melissa (26)(PKU)she gets married in June and is having a 10 day cruise to New Caledonia, Mistry Island & Vanuatu and then 3wks camping in the cape, we'll have to wait till next year to see how it all went, sounds exciting doesn't it. The lovely Stephanie Butler (PKU) was next to arrive, she's an apprentice hair dresser, and her hair looked awesome, I was quite jealous.

Lee Mayfield was next, Lee is an Adult PKU, she has 2 grown up sons 19 & 24. She said she loves apples and lots of vegies, it was great talking to Lee, Lee works for disabilities Qld and she's on 25g protein per day. And that was Townsville clinic.

Whoops forgot Ryan,(22 PKU) he's been in Ireland for 11mths had a fantastic time, is planning a trip to Tokyo and then Canada at the end of the year to go skiing and plans to get a working licence in Canada and work for a while, his brother Zane was not at clinic this year, as he's just started a new job hydraulic hose fitter. A few of us went out to tea at the Yacht Club in Palmer St, Tuesday night it was lovely and relaxing.

We caught the plane at 6.15am Wednesday morning and headed to Cairns, who gets out of bed that early, I'd like to know, the metabolic team were already at the airport. We had a quick cuppa at Townsville cafe and off we go, Viesha 8 (PKU) and her dad Kipp and the rest of the family have just moved to Mareeba, from the gold coast, Viesha who we couldn't get boo out of loves playing the Wii at home, her dad said. Helen, who looked awesome, if I do say so, I nearly didn't recognize her and her lovely daughter Emily. Emily (6)(PKU) loves PKU toast and McDonald's chips, Daniel Dodds snuck in and out, Javan (4)(PKU) & mum Diana, were there, Javan had a play in the inside play room.

Tracey Murray and son Glenn (22, PKU) arrived, Glenn was full of charm, as always, her other son Marc(25) also PKU is now living in Brisbane. Shari Butler is a lovely young lady with a beautiful smile. Baby Jeremy was their, he has Galactosaemia , Stevie(3)(tyrosinaemia type 2) loved drawing and playing in the play room, little miss active. Baby Ella (Propionic Acidaemia),her sister, Mum and Dad where at Cairns clinic, we had a great chat, that was it for Ryan and myself. You blink and the days go so fast…

Thank you to the MDDA for allowing myself and Ryan to go on the far north Qld clinic run to support families where we can, also a big thank you to the metabolic team for having us tag along and thank you to the families for having us there.

Kim Large
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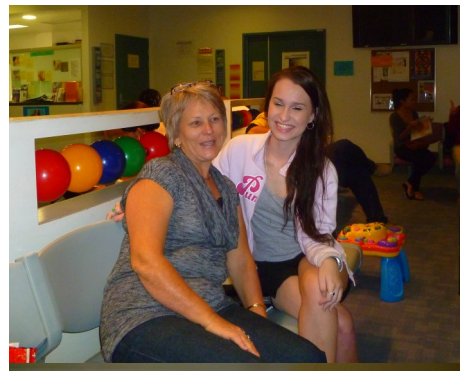

PKU Research at the Children's Hospital at Westmead (Update)

It has been a spectacular few months of fundraising by a small dedicated group of parents and friends, and a special vote of thanks must go to Mirella Nicomede for spearheading a team who staged a wonderful gala dinner, which raised over $130, 00 on behalf of the NSW PKU Association. On top of that, the Rotary Club of Pennant Hills, led by the unflappable Terry Pankhurst, raised another $50,000 all of which has been tagged for PKU Research. We are humbled that so many friends should work so tirelessly on our behalf, and of course the PKU research team at the Children's Hospital at Westmead is spurred on by this faith in us to push our research activites to new levels.

The major focus of our research at present is the further development and validation of our genetically modifed probiotic to treat PKU. Our genetically engineered Lactococcus probiotic bug makes good amounts of the enzyme, phenylalanine ammonia-lyase (PAL), and we have previously shown that in the test tube we get good clearance of phenylalanine using this GM probiotic.

More recently, our Rotary PhD scholar, Naz Al-Hafid, has been testing the robustness of our GM probiotic, which was done by carrying out a series of experiments simulating the environment of the stomach and small intestine, and has shown, at least in the test tube, that our GM probiotic can survive these harsh conditions for the relative short duration of our testing time. Most exciting though was her first experiments in the PKU mouse, where she showed when she feeds them our GM probiotic and labeled phenylalanine, we see a marked reduction in the amount of phenylalanine being absorbed into the mouse's bloodstream. There are many questions though:

1. Will the GM probiotic break down the phenylalanine 'locked' in dietary protein in the gut? Our test tube experiments suggest yes, but we need to do these experiments in the mouse too.

2. Do we need to give the GM probiotic every time the mouse feeds? If not, how many times a day will we need to give the GM probiotic?

3. How long will the GM probiotic survive in the mouse gut?

4. How much GM probiotic will we need to give and how effective will it be in 'real life'?

All of these experiments are planned and will be carried out over the next 12 months.

At the same time, Dr XingZhang Tong, a postdoctural research scientist in our laboratory, who has been working on producing the GM probiotic for about four years now, is working on making an even better version, which will we hope will be even more resistant to the harsh conditions of the small intestine, and which will enhance the functional capacity of the PAL enzyme. He is also planning to explore whether Lactobacillus rather than Lactococcus may be a better way of delivering PAL to the gut.

Naz had an opportunity to present her research at the recent Human Genetics Society of Australasia (HGSA) conference held in Canberra in July, which generated considerable interest indeed from those involved in the management of children and adults with PKU. In fact, she won a prize for her work. Well done Naz!

We're also very pleased to report that Gladys Ho, who has been involved in a number of PKU research project with is over the last four years, will very shortly be submitting her PhD thesis for examination. She too presented her research at the HGSA conference, which was also very well received.

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