Metabolic Dietary Disorders Association

MDDA Matters

Issue #67 September 2018

Connecting worldwide

Two years after representatives from Europe, Canada, United States and Australia first discussed the possibility of an international PKU organisation, and 12 months after the first planning meeting was held, the Global Association for Phenylketonuria (GAP) was officially launched in Atlanta, Georgia on July 5 2018.

MDDA President, Monique Cooper and Vice President, Louise Healy along with Secretary Bianca Albanese and valued member Debbie Colyer OAM, were there to witness this historic event.

Monique and Louise are both proud to be founding trustees of GAP and excited about the future possibilities for PKU. Australia will focus on what can be done to support people with PKU in the South East Asia region. Work on this began on Friday August 10 when Louise met with representatives from the Asia Pacific Economic Cooperation Life Sciences Innovation Forum Rare Diseases Network as part of understanding the status of newborn screening across Asia.

Tobias Hagedorn from ESPKU launched GAP on behalf of the international team who have worked hard over the past 12 months to set up GAP.

GLOBAL ASSOCIATION FOR PKU

Bridging the gap for PKU worldwide

You will see from Tobias' presentation (pg 2) that GAP has ambitious plans for improving the lives of everyone living with PKU around the globe.

The announcement of GAP was greeted with excitement and positivity from everyone present. Already we have been able to identify opportunities for sharing knowledge and resources to provide better support for patients.

We are excited about what this means for the future and look forward to sharing further updates with you as the work progresses.

Bianca Albanese, Debbie Colyer, Monique Cooper, Patricia Guthrie

in this issue

Donations over $2.00 are tax deductible

(Extract from)

Public announcement of the foundation of the Global Association for Phenylketonuria (GAP)

Here is an extract of Tobias S. Hagedorn, Founding Trustee of GAP, speech. A full transcript can be found at www.mdda.org.au

I am Tobias Hagedorn. Since 2001, I am the Secretary of the European Society for Phenylketonuria and Allied Disorders. Since 1997, I am a board member of the German PKU Association DIG PKU. Since 2011, I am their chairman.

My relation to PKU is quite simple, but likely somewhat different than most of yours: I am a husband of a PKU, and together we have two healthy children. So: When I met "my PKU" in 1990, she was sweet little sixteen and she knew exactly about PKU and her diet. I had to learn quickly, first of all that from an outside perspective living with PKU looks much easier than it really is. And I learned how blessed my wife was to be early diagnosed and having access to nutritional therapy.

This is not given all over the world.

I have a dream

I have a dream, that one day all children will have access to newborn screening and early diagnosis, and to adequate treatment. I have that dream!

I have a dream, that one day science will be able to offer us a treatment with minimal impact on our daily life and quality of life. I have that dream!

And I have a dream that PKU patients from all over the world will stand up and call health care providers and politicians to listen to them and to guarantee their dignity.

PKU and Populism

Ladies and Gentlemen, I changed the legendary speach of Dr. Martin Luther King, who was born right here in Atlanta. Dr. King represented a minority that was denied access to essential parts of public life, such as education and equal rights.

PKU patients and their relatives are a minority as well, with limited access to treatment and care, even in Europe and here in the United States of America, which we Germans consider the country of unlimited opportunities. Today, even in western democracies populists become more powerful again with too simple answers on too complex questions. But life is not simple, and we PKU patients and relatives know that better than most of our fellow citizens.

Newborn Screening

Globally, two third of all newborns have no access to screening. Statistically, every hour one unidentified PKU child is born. This means more than 700 PKU patients every month, nearly 9000 every year, all left to the fate of a life with severe mental retardation and physical handicap, that could be prevented by a simple blood spot on a piece of filter paper. Having no access to newborn screening does not only mean to have no access to treatment and care. To deny access to newborn screening means denying equal rights and human dignity to every single patient.

Global Mapping

In most parts of Europe, in Austral-Asia and North America, in all these so-called western democracies and developed industrial nations, health care systems are more or less well organised. We have educated health care providers and trained medical staff. We have industries that supply us with amino acid mixtures and low protein special food. We have researchers who work to collect evidence on the outcome of our treatment and to identify and implement alternative treatment strategies. We have nutritionists and dietitians who help us to make our lives a little bit better every day.

Yes, there are some gaps, even important gaps, such as insufficient reimbursement policies or the different treatment and management guidelines around the world. PKU is the same everywhere, however treatment strategies and access to care is significantly different even in our countries. But we have patients associations and advocacy groups like my ESPKU or your NPKUA and others, who tirelessly work to raise awareness on PKU and call for closing these gaps.

There are regions in the world, where all these achievements are not available, and patients are left behind, undiagnosed and untreated, not even being aware of their hopeless situation. Still, the perspectives of a PKU patient are highly dependent on his or her birthplace. We should not wait for the world to change it. We should take action and make a change ourselves.

Patients impact on PKU care

Patients can make a difference. Let's remember Liv and Dag Egeland, whose mother challenged Professor Følling to

find out the reason for their retardation. Sheila Jones' mother challenged Professor Bickel to find a treatment for her. And Professor Guthrie was motivated by his niece to create a tool to identify early all the few patients amongst all new born.

These are well known cornerstones in the history of PKU, and all of them were initiated by PKU families.

Announcement of the Global Association for Phenylketonuria (GAP)

This week, people from all over the world came to Atlanta to make history in PKU again.

I am proud and deeply humbled that it falls to me to publically announce the foundation of the Global Association for Phenylketonuria as a charitable and independent patients' organisation.

Just to quote another famous American: This is a small step for me, but a giant leap for PKU across the world.

The Mission and Vision

- Our vision is that all people living with PKU despite their origin are able to reach their full potential. We will advance PKU a global health priority, and strengthen PKU associations and groups throughout the world.
- As people living with PKU, we will create a global platform for PKU advocacy that includes ensuring universal access to newborn screening and diagnosis, treatments and comprehensive care;
- We will mentor like-minded patient associations and groups and offer best practices and support that are sensitive to their experiences, culture, languages and laws;
- And we will increase collaboration among PKU researchers, scientists, clinicians and patient groups to move basic science and research forward to accelerate new knowledge, treatments, and a cure.

Human Genetics Society of Australiasia (HGSA) Conference

On Monday August 6, MDDA Vice President, Louise Healy, represented MDDA and Rare Voices Australia at the HGSA conference. Louise spoke to clinical and scientific professionals and industry about a patient perspective on newborn screening and on Rare Voices Australia's plans to advocate for the implementation of the recently endorsed National Newborn Screening Framework.

Louise spoke at a breakfast sponsored by Sanofi, alongside Megan Donnell from the Sanfilippo Children's Foundation and Maddy Collicoat from the Australian Pompe Association. Dr. Kaustuv Bhattacharya facilitated the discussion.

We feel very lucky to work alongside these other rare disease organisations to present a patient voice to those people who are responsible for delivering services to patients with rare conditions. Our presentations prompted some great discussion about the importance of early diagnosis and treatment and about patients working alongside doctors, nurses, scientists and dietitians to get the best outcome for patients.

In addition to the presentation, Louise was joined by Monique and Bianca and we were able to progress the many MDDA projects and liaise with Australasian metabolic allied health staff including nurses, dietitians and nutritionist about several key MDDA wellbeing and patient support initiatives we are collaborating with the clinicians on. We were able to have meetings with the metabolic dietitians and the ASIEM executive team members. We also had a really interesting meeting with an organisation that are managing a clinical trial in Australia for a PKU treatment – we will discuss more about this at the retreat.

MDDA are grateful to Sanofi for providing an opportunity to present a patient voice at this conference and excited that we had the opportunity to engage with our clinicians to further key MDDA initiatives.

Why your membership matters

Membership ensures MDDA can continue to support all individuals living with an IEM and help them achieve positive health outcomes

- provide more informed choices, resources and support
- provide support by way of website, newsletters, recipe resources, Peer support, conferences, family retreats, social activities and many other programs.
- enables MDDA to provide a voice to government and decision makers on all levels, to promote research into inborn errors of metabolism
- to raise awareness to the greater community and to advocate for all individuals and families living with an IEM.

Please contact us office@mdda.org.au if you would like to check on the status of your annual subscription or would like to become a member of the MDDA.

Not going this year? Dont worry a full write up on our retreat will be in our end of year newsletter. Information from the sessions will be made available to members.

NPKUA 2018 Conference

After a whirlwind trip across the US, it was only once I had landed back on home soil that I was able to truly appreciate the experiences I had at the NPKUA conference.

Over the course of the conference, I met so many amazing people, all with different connections to PKU.

PKU adults. We live different lives and had very different upbringings, yet we go through the same challenges every day. We all connected instantly and the support we shared is unparalleled. I love that I have been able to finally put faces to names, being online Instagram friends for years, and have transformed that online bond into one that will last a lifetime.

Parents of PKU Children. I met with parents of little ones with PKU. It was a privilege to pass on my knowledge about what it was like growing up with PKU; travelling, studying and just generally living while sticking to diet. PKU has not been a barrier to my aspirations and I believe anyone should shoot for whatever they dream.

Medical Professionals. I met with researchers developing emerging treatments, medical professionals and medical food representatives (and tasted some amazing samples!). I learned so much from a scientific standpoint which I'm grateful for. Ten years ago, there was only one company in the PKU drug development space. Now, there are nearly 20 companies working toward PKU drug developments. It was great to see what is in the pipeline for the years to come.

A definite highlight was meeting Dr. Geir Folling, the grandson of Dr. Asbjorn Folling, the man who discovered PKU in 1934. Dr. Geir presented the incredible story of how his grandfather's discovery came to be. Dr. Folling survived tuberculosis and a near miss with a train to go on to be the first to publish the link between PKU and mental retardation. To think, had events panned out differently in Dr. Folling's life, we and the many I had met that weekend at the 2018 NPKUA Conference would be far from the successes we share.

I was also extremely lucky to have had the opportunity to sit down with Jack Everitt, an English PKU adult & his friend Liberty in Atlanta to film for their upcoming production "So what can you eat?". Being born with PKU has thrust us into a world where nutritional awareness is at the forefront of our minds; every second of every day. Our efforts are somewhat invisible though, as our day to day treatment avoids something horrible, brain damage. To the outside world we are normal, however our dedication and persistence is truly highlighted when our lives are starkly contrasted with that of a non-treated PKU individual. This film documents an experiment by Liberty (non-PKU) who takes on PKU dietary therapy to highlight not only its difficulty but also challenge society's relationship with food. Keep your eyes peeled for this! (Coming to Netflix!)

I was also left quite humbled, coming to the realisation of how lucky we are in Australia. PKU has no borders and doesn't discriminate, however a person's country of origin and residence does determine the quality of care received as a PKU patient. In the US, a country that is pioneering the first pharmaceutical treatments for PKU, a gap still exists in basic care through the lack of access to medical foods and formula. Not to mention other countries where newborn screening simply does not exist. The conference played a great base for international discussion on how initiatives can help bridge these gaps worldwide.

The power of being in a room surrounded by hundreds of faces, from different places, who may seem like strangers, but felt like family, is an amazing feeling. Everyone just gets it.

The strength of the PKU community worldwide is a true testament to the various groups that exist in every corner of the globe. Thank you to NPKUA for the 2018 Conference Scholarship that assisted me in attending.

I am so glad to be part of this along with Monique and Louise (who were fantastic travel buddies may I add!). The unveiling of GAP, the Global Association for PKU, is a united extension of this, to work toward greater care for those living with PKU worldwide. Exciting times ahead!

Bianca Albanese

Diet for Life Trudy's story

My name is Trudy Smith. I was born in August 1964 in Brisbane. At that time the only test available for PKU was the nappy test, but it came back negative. Everything came back normal. However, my mother felt there was something not quite right, so kept taking me back to the doctor. The doctor told her that she was worrying about nothing and being too fussy and that she should take me home and relax because there was nothing wrong.

When I was 10 ½ months old I started to have convulsions so my mother took me to a naturopath who told her that he didn't know what was wrong but was sure it was connected with my food. He made arrangements for me to see a paediatrician. After many tests it was discovered that I had phenylketonuria. The doctors realised that my diet had caused damage to the learning part of my brain and that my parents should consider putting me into an institution because they felt that I would never be able to walk or talk or be able to live a normal life. My parents were shocked and devastated but dug their heels in and said no way, and so my journey with the PKU diet and formula began.

Cynogram was the formula at that time which had the look and texture of grey wet cement. Each meal was a nightmare for all of us as I would try to fight off having it. It was foul. I would no sooner get some down, then vomit most of it back, and then mum would have to start all over again. It took about 18 months before I could take it without bringing it back up. Even now when I smell soy sauce it reminds me of the taste of cynogram. I also had a calcium tablet every day to help with bone growth. After a few years the formula was changed to aminagram which was a bit more pleasant.

Through the Lions club for a few years at Christmas time, my mother would take me to Melbourne for the PKU Christmas party arranged by the chief dietician Mrs Betty Lynch. It was good to see other kids eating the same sort of food as I was able to eat. In 1966 my brother Gavin was born but tests proved that he was PKU free. Gavin was my best friend and we are still very close today.

I went off the diet by the age of 12, as, at that time it was believed you didn't need the special diet anymore. I attended special school until I was 16, then worked in sheltered employment for some years. I enjoyed the physical work, especially working with timber. During this time I obtained my forklift licence which I was very proud of. Then there was a change and I worked as a cleaner in the Lutheran book shop and also enjoyed making and painting pottery.

As I was getting older I used to find I would quite often run out of energy especially during the afternoon. Around this time we heard about a young woman who had PKU and was still on the special diet. After many enquires I got an appointment with Dr Bowling at the Mater hospital in Brisbane. During this appointment it was discovered that all my records and files from the Royal Brisbane Children's hospital had disappeared.

It was as if I had never been there so I had never been contacted regarding the need for me to return to the PKU diet and formula. Dr Bowling was very thorough and it was recommended that I resume the PKU diet and formula. He also added Tyrosine to my medication. After a while I began to feel as if I had more energy and things became a little easier for me. The formula at that time was PKU Express. That was much more palatable and easier to take.

We were attending a church in Queensland and I became interested in the bass guitar. It was quite a challenge for me but with a lot of support and some lessons with a very patient and understanding teacher, I was able to take my place in the music team at church. This gave me a feeling of great achievement. I believe that my faith in God, and in Jesus Christ, has helped me to overcome the difficult situations in my life.

In 2006 we moved to Lakes Entrance in Victoria. My local doctor had never had anything to do with anyone with PKU so it was quite a learning curve for him at that time. I was referred to the team at the children's hospital in Melbourne where, during my first visit, some of my missing records were found. It was recommended that I have yearly visits to the hospital, which meant a four hour drive each way, and to continue with monthly finger prick blood tests to monitor my levels. Since that time with strict attention to my diet my levels are generally pretty good. After a time the clinic moved to the Monash medical centre at Clayton and then back to the Royal Melbourne Hospital where I am under the care of Dr Julie Panetta and her team. They have been and continue to be a great support to me.

I now work as a volunteer two half days a week at one of our local opportunity shops where my main task is sorting and steaming the never ending supply of donated clothing. I find my levels of concentration can go astray sometimes but on the whole I am able to manage with the help of family, especially my mum. I am interested in many areas of craft and enjoy things like cross stitch and tapestry in my spare time. I live with my mother and step father and we are all involved with our local Church. We have two small dogs who are great companions. During the last few years we have been able to take a few cruises to many different places including New Zealand, Tahiti, and Hawaii. These cruises have been awesome especially as, with my restricted diet, most of my meals are made to my special needs.

While it can sometimes be rather frustrating, especially when eating out, at the limited types of food that I am able to enjoy, I know that it is mainly due to the low protein foods that I can have that make it possible for me to enjoy the life that I now have. I agree with the title of the article by Con Giannas which said, "Diet For Life".

Trudy Smith's story as shared by John & Shirley Ray.

Young adults living with an IEM

OTC

Brooke Hall

Age: 20 IEM: Ornithine Transcarbamylase (OTC)

Describe your family & growing up with OTC:

I've grown up and lived with my mum Trish, who is a carrier of the OTC deficiency gene and my sister Georgia who is completely free of OTC.

Growing up with the condition to me, was like being a normal kid. Being born with OTC, I didn't know any different than taking my medication, hospital admissions and a low protein diet – which I guess in the long run has been a positive.

How do you explain OTC:

I always tell people that it's a genetic disorder and its definitely not contagious. My body is missing an enzyme in my urea cycle that breaks down ammonia. I then explain that when you eat foods that have protein in it, it breaks down into ammonia and my body is just missing one step to help it break down, so I take medication to help get rid of it and eat low protein foods so I can reduce the risk! I always mention that too much ammonia is deadly in your body as well!

How were you diagnosed?

I was diagnosed via an amniocentesis when my mum was pregnant, before I was born my mum lost a child at 5 days old because of undiagnosed OTC.

What is the treatment:

My treatment is taking 6x Citrulline tablets 3x a day, and eating low protein foods. Often sticking to around 7-8g of protein a meal. If I do end up hospitalised, the treatment is intravenous Sodium Benzoate and Arginine until my ammonia comes down.

What are the challenges you/ family have faced from the point of diagnosis to now?

I think that adjusting to changes has been difficult, countless hospital admission means putting mine and my family's life on hold. I think it's more difficult for family rather than myself sometimes!

How do you go about educating family and friends?

I am totally open to explaining my condition to family and friends, I find that I'm happy to talk about it and explain it. Also, I find that not hiding things on social media and in life makes things a heap easier. I guess when people can actually see what's happening, they are more likely to understand.

What is your favourite thing to do to relax?

I love sport!!! But I guess that's not relaxing, I do competitive sport aerobics and netball. When I'm not doing that or working at an indoor trampoline and laser tag park, I love watching reality TV and playing with my dog!

Describe a regular dinner time?

I often cook my own dinners and cook heaps of vegetables! I usually have a Vege Pattie, or curry, stir fry, pasta or a small amount of white meat (Pork, Chicken or Fish).

What is your favourite food?

Mushrooms, Pumpkin, Asparagus, Hot chips and any type of creamy pasta!

How do you deal with food when travelling/going out?

I always take my own snacks and basics when travelling, other than that when going out, I usually go for some sort of vegetable risotto or something I know is pretty low protein and if I'm unsure, I just ask!

What did the teachers/school/ workplace know about your condition?

My school I went to knew about my condition and were really quick to action when I went unconscious with a high ammonia at school.

My workplace knows about my condition, but I didn't tell them until after I was hired, because I believe OTC shouldn't stop me from doing a good job at work.

How is the workplace in accommodating any special requests/requirements?

My workplace is amazing with my condition. They let me take my medication when needed and eat when I need to. After informing management about my condition they took everything into consideration – and really understand when I have to be in hospitals to help my ammonia come down.

What would you like everyone to know about growing up/living with OTC?

I definitely think you can't live your life in fear that something bad will happen. For a while, I was fearful to go to restaurants or go anywhere without some sort of safety net (My mum for example) in case something was to happen. But as I've grown up I've learnt that I know my body better than anyone else does, and I am in charge of my diet and medication.

Any tips for parents or kids living with OTC or an IEM and ensuring it is as easy as possible?

Whatever works for you. I think planning ahead if your travelling or going out is key to assist you're diet and medications.

Try not to make a huge fuss out of it, in social situations like calling out that the person with OTC has to eat something different!

..... and don't let it stop you, you can totally live your best life with OTC and achieve ANYTHING you want to achieve!

Elena Stahlhut

Age: 18, IEM: Phenylketonuria (PKU)

My name is Elena and I tuned 18 in July this year. There are 4 people in my immediate family. I have an older sister named Madison who is 21. My Mum and Dad's names are Ingrid and Scott.

How were you diagnosed? As a baby, I had the heel prick test which tests for disorders such as PKU, which showed up as positive in my test. The doctors explained to my parents and myself also, when I was older, that each parent has a 'good' gene and a 'bad' gene. Unfortunately, I got the 'bad' gene from both parents which resulted in PKU. That is how we found out that I had classical PKU.

What is the treatment? There is no cure for classical PKU yet, that I am aware of. However, I must take a liquid supplement 3 times a day to keep my Phenylalanine levels as low as possible, and to stick to a low protein diet to keep my body healthy and functioning properly. I used to be on a supplement called Maximum and when they brought out new and easy to use supplements, I would give them a try. It took me a total of 5 or 6 years to change over supplement as I didn't like most of them. I now take Lophlex Orange which I have with apple juice to make it not taste so nasty. Nutricia and other PKU friendly brands are making my life easier with these new supplements and foods, they just take a bit of time to adjust to the change.

What are the challenges you/family have faced from the point of diagnosis to now? Some challenges my family have faced have been when I was younger I once refused to take my drink. My Dad talked to me about it and I decided to take my drink again. I was 4. I think going on primary school camps were challenging for my family. Trying to make sure the teachers on camp were aware of what I could and couldn't have to eat was challenging as they had so many children to look after that they didn't really have time to check what I was having. I try not to let my PKU diagnosis rule my life. Obviously, it's a big part, but I don't let it stop me from doing what I set out to do.

How do you go about education friends and family? To be completely honest, if I am at University or somewhere social where I am meeting new people, I will tell them I am a strict vegan and that's my diet that I cannot waver from. Just from personal experience, I do this as I find it easier for people without a metabolic disorder to understand. However, all my close friends and family know all about my condition and are very supportive about it all.

What is your favourite thing to do? My favourite thing to do is to dance, listen to music or going out with my friends.

What would a menu for a day look like for you? Meal times for me are usually fun and exciting as I love food but if I'm eating out it can be difficult. What I eat in a day would usually be, a smoothie or avocado toast for breakfast. A salad or wrap for lunch. Snacks throughout the day would be veggie chips (yum) or fruit or crackers. And for dinner My mum, Dad and I always look in cook books or the internet for new recipes. My dad trained to be a Chef, so dinner time is always exciting.

GROWING UP – SCHOOL. At primary school, all my teachers knew about what I could and couldn't eat as I was much younger and a little more reliant on adults. For excursions and parties my Mum being the best Mum ever would always pack me the foods I can eat so that I never felt left out. Because of that I always had a little cupcake for someone's birthday which was great!

Describe the transition into secondary school, your teen years and becoming more responsible for your PKU: When I entered high school the only adults that knew about my disorder was my PC (Pastoral Care) teacher, who is the teacher I check in with every day (morning and afternoon) as well as the school nurse. This gave me a bit of freedom to be more responsible and realise how responsible I will need to be with my diet and what I am eating. During high school when parties were on, I will admit I may have given into temptation maybe 3 times in those 5 years, but I would always make sure I told Mum, so it could be sorted out later. I didn't take in my own cupcakes for birthday parties as I guess I grew out of it. I didn't really mind that I wasn't eating what everyone else was. My PC teacher always made sure there were options for me to eat anyway, so all was good.

Tell us about your work: I have 3 jobs in total. I work as a dance teacher for levels 6 & 7 Jazz at Bluerhythmix Dance School in Highfields. I also have 2 retail jobs at Valleygirl and Temt in Toowoomba. The dance school owner is a family friend and used to be my Daycare Mum so she knows about all about PKU. I haven't told my Managers of the retails jobs as it's not something that would affect my work. I also have a side business doing makeup for parties, formals and other special events.

What is your favourite part of your job and why? My favourite part about teaching is that I'm teaching my students dance, which is my biggest passion. I just enjoy every moment I'm at the studio. And my favourite part of my retail jobs are the girls I'm working with, they make work fun.

What are your hobbies/interests: Dance, music, makeup, skate boarding, going to the gym, adventuring with my friends and going out with my friends.

How do you plan for going out/away? When I go out to eat, I always google the place, for example a restaurant, and I look at the menu first to make sure I can eat there. My friends are always very chilled about where we go because they're so supportive and don't want me to be left out. So if it's somewhere that I can't eat, then we'll go somewhere else that is better suited. For holidays, when we go away, Mum and I make lists of things to take with us, such as low protein foods and regular foods that are suitable. We then plan what I eat in those days to make sure nothing goes wrong and that we don't have to worry about getting to the end of the day and having to make up my protein intake or having something completely free as I'm up to my total for the day.

What would you like everyone to know about growing up with a metabolic disorder? Its hard and very frustrating sometimes. I always got annoyed that I couldn't eat like a normal person and it still vexes me sometimes, but I do get over it. Sometimes the food isn't the only problem. Sometimes mental health or our moods go out of whack and we need a rock to bring us back down. Which is why it is good to have a great support system like a family.

Any tips for young adults/teens? I think connecting with other people who have the same disorder can really be beneficial because it shows you that you are not alone. Mum is always my best contact to rant too but it is also nice to be able to connect to someone the same age with the same experiences. Also, I've learnt not to let your friends have a sip of your supplements. "It can't be that bad!" It is and its hard to get vomit out of a carpet. Woops!!

Any extra comments/experiences that you wish to share

1. I wouldn't be here, alive and healthy without my gorgeous mother
2. If anyone is around my age and would like to connect, you can find me on the MDDA Facebook page!
3. Dr Jim and Anita are the best!

FUNDRAISING UPDATES

Cut for a Cure

The goal: $5,000 to be raised from an uncle cutting off his dreadlocks he had for 11 years

The result: $15,530.76

Ashlee McGill, a MDDA member, contacted Monique in late March to discuss a potential fundraiser idea she was floating after having participated in the Great Protein Challenge.

Ashlee has 2 children, Kaiden (4) and Alissa (1) who were born with PKU, and their uncle "Skip"- Justin, who has had his dreadlocks for 11+ years knew it was time to cut them off...

"Everyone has always wanted to cut a dreadlock over the years – well, how better to raise the money, for my nephew and niece who have to struggle with the challenges of PKU every day. This is the smallest price I can pay compared to what the McGill, and many other, families have to go through every day. It was time for all those people to put their money where their mouth was" Justin said.

Prior to the event, the MDDA and Ashlee, worked closely together to create a portal where donators were directed to the MDDA page to "purchase" a dreadlock or 2, or 3, or 4, and get some more insight into what PKU is and why the MDDA was chosen for these funds.

'Monique, Jenny and the team at the MDDA were excited to get on board and assist us in any way they could to make this fundraiser a reality and a success" Ashlee said. "Their excitement and support were a motivation".

As the event drew nearer, the dreadlocks were being purchased in truckloads and the reach for the event spread to many PKU families. "I was receiving messages of support from other PKU families that were grateful for what we were doing to raise PKU awareness and it sunk in that this wasn't just about our family anymore – it was bigger than that! The support from other PKU families pushed us to make this event as big and as successful as we could for a 'hair cut'" Ashlee commented.

And so, the idea became a reality.

VIC

On Sunday 27th May 2018, family, friends, colleagues, strangers, all joined Ashlee McGill at the Cardinia Golf Club in Beaconsfield Upper, Victoria, for an afternoon of family fun... and the big cut!

Over 150 people attended the event on the day, with Monique – president of the MDDA also showing her support and taking the time to explain the MDDA's involvement with PKU. Monique also spent her time chatting to family and friends – 'It was great to talk to Monique about the effects of a PKU diagnosis on the extended family', Bianca – Ashlee's sister and 'Skips' wife said. "As a mother myself, and a big sister to Ashlee, I feel I have an inbuilt sense of protection for Ashlee. The diagnosis of Kaiden, firstly, having PKU had a knock-on effect because as her family – I wanted to fix everything and make everything better! And this time... I couldn't!! Extended family ride this wave with the parents of PKU and although not directly affected by the daily challenges, we also make lifestyle changes in whatever way we can to support. This is the exact reason that my husband wanted to do 'something' to support Kaiden & Alissa, because we are on this wave with them".

"Our 'Cut for a Cure' event was a huge success and it was such a humbling experience to have so many family & friends get involved, one we will never forget" said Ashlee.

"I've said many times but will keep on saying, we will forever be grateful to Skip for his sacrifice towards PKU and giving us the ability to make our first fundraiser a reality with such huge generosity. The final amount was $15,530.76 which has completely blown us away!!

I dream of the day being a reality, where my kids will be able to live a life free of the many challenges that PKU brings." Ashlee McGill.

MDDA thank Ashlee, Justin, Bianca and all their family and friends for this amazing effort.

Yarra Glen Lawn bowls tournament

Thanks to Sonia and Peter Hellings who took part in the Yarra Glen community bowls tournament, over 12 weeks, representing the MDDA and Yarra Flats Baking Co. Thank you for nominating MDDA as your charity and for the donation of $250.

Ironbody Crossfit Gym

WA

Thanks to member, Felicity Simmons, from WA who organised fundraising through her local gym and raised $670

TAS

Weighing in at a touch over 158kg Brett decided that it was time to look after his own health and shed a few kilos. He also wanted to help make a difference in the lives of the Tasmanian Children living with PKU. This was the start of putting together "Egg's 8 Week Weight Loss Challenge" where people were able to pledge a dollar amount donation for every kg that he would lose across the 8 weeks.

Egg received huge support from the local community with people pledging donations from $1 per kg through to $10 per kg, one pledger donating an extra $100 on top of his $10 per kg donation if Brett was able to lose over 30Kg.

After the 8 weeks of dieting and moderately exercising Egg had lost a whopping 28kg. Brett said "having the donation challenge in the background was a big help in keeping focused'. So much so that he didn't deviate from his regime even once.

A massive thank you to Brett and also Lauren and Justin Penneyston who have driven Tasmania's fundraising efforts.

TAS: upcoming

QLD

Fun run

On the 16th September Bianca Albanese, a PKU adult, will be running the Blackmores 10km Bridge Run. This run is a catalyst for wellness within the community, both through an individual's participation and it's platform to raise money for charity. Bianca has chosen to donate all funds raised to the MDDA, where she holds the position of Secretary. Since beginning this role, Bianca has been inspired by the amazing work the MDDA team achieve, advocating for individuals and their families living with an Inborn Error of Metabolism. Your donation will make a great difference to the lives of those living with these rare genetic disorders. Please get behind her and help her reach her target by donating! Go to **givenow.com.au/mdda**

ideas

Have a look at the MDDA website to see our new programs and research initiatives for 2018/2019.
Get involved today in fundraising-

Approach your local community club - Rotary/Lions

Local Sausage sizzles

School/work gold coin donations for free dress days

Fun runs

Call the MDDA office and we can help you get started TODAY!

Far North Queensland Metabolic Clinics

The Far North Queensland Clinics were once again held in Townsville and Cairns during May. These outreach clinics are an important medical and social date in the calender for our IEM community in these areas.

Thanks to Kim and Roy Large for taking time to connect with new families, catch up with familiar faces and educate about the support, vision and goals of the MDDA. Kim and Roy have twin adult sons with PKU, and are always offering to help with these clinics and any get togethers in Far North Queensland. They spent a week driving from Proserpine to Townsville, then Cairns before heading back home. Their knowledge is key in helping to foster the importance of attending the clinics amongst new families and also spreading their love of clinic that they were fortunate to experience with their boys. The importance of meeting and connecting with other families in similar situations is invaluable.

Cairns

Townsville

PKUNSW Updates

12 August – City2Surf

Another year has passed and the PKU Association of NSW once again took part in one of Australia's most renowned road races - Sydney's City2Surf. The City2Surf is a 14km route from Sydney CBD to Bondi Beach with an estimated 80,000 competitors who participated. Well done to all who participated.

2 to 6 October PKU Youth Camp

We wish all those heading off to the youth camp at Berry in NSW a fantastic time. It is a great opportunity to learn about how others manage their PKU, become aware of other food ideas whilst building confidence, problem solving skills and making lifelong friends. Contact PKUNSW for more information on current and future Youth camps or go to **pkunsw.org.au**

2 June - Gala Dinner

The PKU Gala Dinner was held on the 2nd June. Congratulations to all involved. What a fantastic result, raising $156,000 to go to PKU research.

**calculate all recipes as per guided by your dietitian*

Adapted from "Apples to Zucchini"

Sicilian Dip

Ingredients:
2 teaspoons olive oil
1/2 medium onion, chopped
2 large cloves garlic, minced
1 cup canned crushed tomatoes
1 teaspoon lemon juice
5 sun-dried tomatoes coarsely chopped
4 large artichoke hearts
1/4 cup pitted green olives
1/4 cup chopped fresh basil
2 tablespoon fresh chopped parsley
salt and pepper

Method:
Heat oil in fry pan. Add onion and sauté until soft Add garlic, then stir in tomatoes and lemon juice. Bring to a simmer.

Simmer for 2 minutes, stirring. Remove from heat and set aside.

Combine sun-dried tomatoes, artichoke hearts, olives, basil and parsley in a food processor. Pulse until vegetables are finely chopped. Transfer to medium bowl. Stir in cooled tomato mixture and season to taste with salt and pepper.

Serve with assorted cut raw vegetables.

Makes 2 cups

Grilled Potato Salad with Lemon and Herbs

Ingredients:
Dressing:
1 tablespoon Dijon mustard
Grated zest of 1 lemon
2 1/2 tablespoons fresh lemon juice
1 minced garlic clove
1/2 cup olive oil
salt & pepper
Salad:
1kg baby potatoes
3 tablespoon olive oil
salt & pepper
4 spring onions
1 tablespoon fresh parsley
1 tablespoon fresh thyme
1 tablespoon fresh oregano

Method:
Dressing: Place all dressing ingredients in a bowl and whisk together. Season to taste.

Salad: Boil potatoes for 7 minutes drain and set aside to cool.

Toss the potatoes with oil and salt and pepper. Place on grill and grill over medium until fork-tender (approx 15mins) turning every 5 minutes.

Let cool slightly then cut each into half.

In a large bowl, very gently toss potatoes with the dressing, onions and chopped fresh herbs.

Serve and enjoy!

Zucchini and Tomato Casserole

Ingredients:
2 Tablespoons olive oil
1 large onion quartered and thinly sliced
2 medium cloves garlic sliced
1 1/2 teaspoons chopped fresh rosemary
2 thinly sliced fresh sage leaves
1 teaspoon chopped fresh thyme
salt & pepper to taste
420 grams zucchini cut into thick slices
2 large tomatoes cut into thick slices
8 pitted black olives quartered

Method
Preheat oven to 175 degrees. Lightly oil a baking dish and set aside.

In a large fry pan heat 1 tablespoon of oil then add onion, garlic, half of the herbs and several pinches of salt. Reduce heat to medium low, stew the vegetables gently for 5 minutes. Remove from the heat and spread the onion mixture evenly over the bottom of the baking dish; season to taste.

In the same fry pan add zucchini, slat and the rest of the herbs. Sauté, stirring frequently, until zucchini turns brown. Remove and spread over the onion mixture. Spoon the tomatoes throughout the casserole and sprinkle the olives on top.

Cover with aluminium foil and bake for 25minutes. Let the dish rest for 5 minutes then serve.

Makes 5 cups

Product update

Obap: Sweet potato noodles

Protein per 100g: 0.1g
Protein per serve:0.05g
Serving size:50g
Available from:
Woolworths

Culinary Choice: Cheese & crackers

Protein per 100g: 1.8g
Protein per serve:0.9g
Serving size: 50g
Available from:
Coles & Woolworths

Eden Pantry: Organic Coconut icecream

Protein per serve: 1g
Serving size: 75g
Available from:
Vegan Essentials &
Healthy Being stores

Arlington: Tapioca pudding-Mango

Protein per 100g:0.4g
Protein per serve:0.4g
Serving size: 110g
Available from:
Woolworths

Bell Farms: Bubble 'n Squeak

Protein per 100g:2.2g
Protein per serve:1.6g
Serving size:75g - 1 pattie
Available from:
Woolworths

Zero: Fettuccini

Protein per 100g: 1g
Protein per serve:1g
Available from:
Woolworths

Always check the nutritional panel on products

This newsletter will only grow and get better with your input.

Please share your stories, ideas and tips, birthday celebrations and functions with us via office@mdda.org.au

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The MDDA is supported by funding from the Australian Government under the Health Peak and Advisory Bodies Program.