

Metabolic Dietary Disorders Association

MDDA Matters

Issue #68- December 2018



“Embracing the Potential”

We can do things we don't even dream we can do...when you embrace your potential you acknowledge the small and large contributions you make, listen to and respect yourself, seek balance and have the courage to live your life to the full.

Embracing the Potential was the theme at this year's retreat. It's about identifying, realising and embracing your journey of personal growth and uncovering the greatness that lies within.

Over 140 members embraced the weekend with many commenting how they found the retreat to be so inclusive and supportive. It takes a lot of time and hard work organising the retreat, making sure we provide the right mix of education, workshops, interesting speakers, valuable social time and finding the right place to cater for our group.

It is opportune that we recognise two members who go above and beyond their voluntary roles, **Monique Cooper** and **Louise Healy** who willingly dedicate their time and energy to MDDA. They are both parents that are not only invested in the best outcome for their own children but for every individual living with an IEM. Their scope of work goes far beyond the roles of President and Vice President.

Monique and Louise's commitment, enthusiasm and positive approach to ensure a great result for all MDDA

members and the greater IEM community has ensured MDDA has continued to successfully execute on our association's vision, mission and goals.

They continually exceed expectations of their voluntary positions offering advice and guidance, developing and progressing initiatives, maintaining professionalism when dealing with all stakeholders, government and members, always seeking the best possible outcome for our association.

Much of what they do goes unrecognised, receiving and making phone calls and emails giving the IEM community a voice to government departments, parliamentary engagements, reviewing government health policies and programmes, submissions, consultations and IEM education with the wider health sector, international representation ensuring the IEM community has a voice. At many times putting aside their own businesses, personal and family life to ensure our voices are heard. MDDA are very fortunate to be beneficiaries of Monique and Louise's commitment.

Thank you, Monique and Louise, for always going above and beyond for us.

We wish you a safe and happy holiday season – Merry Christmas and Happy New Year! We look forward to sharing 2019 with you all.

Jenny and Kerry

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Metabolic
Dietary
Disorders
Association

Rare Voices Summit 2018

MDDA were pleased to attend Rare Voices Australia "Rare Disease Summit" in Melbourne 16-17 November.

The summit opened with an announcement from The Hon. Greg Hunt MP, Minister for Health on the Morrison Government's support of a National Rare Disease Framework. Congratulations to Rare Voices Australia who have successfully advocated for the 2 million Australians living with a rare disease to promote for an effective rare disease policy.

The summit provided MDDA with an opportunity for networking with representatives from other rare disease support groups along with clinicians, key government, scientific and pharmaceutical stakeholders. It is amazing to think how far we have come from just 3 years ago when we last attended this same summit and the extent of advocacy work and strategic relationships MDDA has built over this time. It is so important to bond together as a rare disease community and ensure our voice is heard loud and clear, to raise the profile of rare diseases and be part of guiding rare disease policy.



The two-day event was a mix of plenary sessions and breakout workshops with Rare Voice's objectives being:

- To facilitate broader stakeholder engagement and collaboration between patient organisations, the Government, clinicians, researchers and the pharmaceutical industry on important issues in rare disease.
- To strengthen the rare disease voice in Australia.
- To highlight examples of best practice, policy reform and improved patient outcomes.
- To confirm the role and unique positioning of Rare Voices Australia: patient centred, with demonstrated experience in working with all stakeholders.

You can read more on the National Rare Disease Framework below.



Australia's first National Rare Diseases Framework

Media Release 16th November 2018

The Australian Government announces they are taking more action to support people battling rare conditions by developing Australia's first National Rare Diseases Framework and Action Plan.

The Liberal National Government is taking more action to support people battling rare conditions by developing Australia's first National Rare Diseases Framework and Action Plan.

Funding of \$170,000 will be provided to Rare Voices Australia through the Government's Public Health and Chronic Disease Program to enable the collaborative development of the action plan and framework.

Specific priorities, actions and activities will be identified through extensive consultation including people with a rare disease, clinical and academic experts, policy makers and state and territory governments.

The framework and all recommendations for action will be supported by robust evidence so that people with rare diseases can receive timely diagnosis, treatment and the best care possible.

Our Government recognises the fact that these diseases are statistically rare – with an estimated prevalence of five in 10,000 – therefore special and concerted efforts are needed to address them.

There are more than 8,000 rare diseases which are life-threatening or chronically debilitating.

When someone is diagnosed with a rare disease they need our love and support but also the best possible treatment and care. This plan will benefit people with rare diseases, their families and the wider community.

The Government provides treatment for rare diseases through Medicare, through medicines subsidised through the Pharmaceutical Benefits Scheme (PBS) and the Life Saving Drugs Program.

Already, through the Life Saving Drugs Program, the Government funds fourteen different life-saving medicines for nine very rare diseases providing physical, emotional and financial relief for 400 Australian patients.

The Liberal National Government's strong economic management means we can also support medical research that gives Australians with rare diseases access to the next generation of life saving technologies.

Authorised by Greg Hunt MP, Liberal Party of Australia, Somerville, Victoria.

Rare Voices Australia response to the announcement can be read online at rarevoices.org.au

Future Genetic Counsellors learn from MDDA experience

Hi! We're Gen and Renee, you may have met us at the 2018 Retreat in Adelaide. We're both Genetic Counselling students and have just completed the first year of our master's course. As a part of the course we had to complete a community placement and we were lucky enough to be placed with MDDA!

Outside of our course we both have very similar interests. With a love for keeping fit and exploring the outdoors, going on hikes. Gen knows some great local spots and Renee's favourite hiking spots are all in the blue mountains (she's a Sydney local). We both also love food and cooking and enjoy experimenting with different gluten free and vegan options.

We both have a background in biological sciences and have a great love for animals. Starting out our undergraduate degrees we were both interested in conservation. We soon, however, realised a love for genetics and working with people over animals or in the laboratory. This led us to genetic counselling!

A genetic counsellor provides education and support as well as options for genetic testing for people with a family history of a genetic condition. During our placement, we discovered that many people with an IEM do not see a genetic counsellor. We believe this is due to the management through diet, with the genetic nature of the condition making no difference to treatment.

We really enjoyed our time at MDDA helping the association out with a variety of different tasks in the office. We then were lucky enough that the association supported us to head along to the 2018 retreat in Adelaide! This was an amazing weekend and it was great to get to meet so many of you!



Renee & Genevieve enjoying the retreat photo booth!!!

Throughout the retreat we helped out behind the scenes with Kylie making sure everything ran smoothly for you all. We also did a lot of work with the kid's club and had a fun time helping them go shopping and create their beautiful cupcakes! We also helped record the event getting lots of photos and videos that I'm sure you will see in coming newsletters and in the Facebook group!

Another great component of the retreat for us was speaking with you all and hearing about your personal stories living with an IEM. Hearing personally from you all was a great experience and allowed us to gain a great understanding of what it is like to live with an IEM. We were able to take away so much from these discussions to put into our everyday work as genetic counsellors in the future.

Overall our experience with MDDA was great and we have learnt so much from you all! Thank you so much to everyone who shared their stories with us and made us feel so welcome within the MDDA community! Thank you as well to MDDA for hosting us and supporting us to go to the retreat in Adelaide!

How Low can you Go?
the greatproteinchallenge.com.au

THE GREAT PROTEIN CHALLENGE
FEB 2019

The great protein challenge is back again in February 2019.
Play by yourself or register a team and help each other reach the goal of eating under 10 grams of protein for just one day.

SA Family Retreat



“Embracing the Potential”

Adelaide Hills Convention Centre – 21-23 September 2018

Monique Cooper, President opened the SA MDDA family retreat to over 140 members eager and ready to meet, network and connect in a relaxed and social environment while furthering their knowledge and shared experiences of living with, or caring for someone with an IEM.

Attending your first retreat can be somewhat daunting but also a significant life changing experience - especially if you have never met anyone before who shares your story. Monique welcomed all and encouraged us to open ourselves to new opportunities, information and friendships – participate actively in discussions and seek out new allies to walk alongside in our IEM journey.

MDDA believes wholeheartedly in a future where ALL individuals living with an IEM are leading a life at full potential, not limited by choice or resource. It is what drives every initiative we plan, every campaign we work on and every conversation we start or have – whether that be in a political, social or closed environment.

This retreat weekend was just another step closer to us realising this dream - through not only providing more choices and unveiling more resources to our IEM community, but also sharing exciting new progress being made on the IEM and PKU research front, that opens possibilities and gives hope for a brighter future for all IEM Australians.

This weekend encouraged us to pursue a journey of new information and enlightenment, and in doing so, many discovered the potential to embrace ourselves.

The following is a synopsis of the weekend program;

Friday 21

On the shoulders of Giants - How PKU/IEM families have shaped history and PKU/IEM Global Perspectives and Priorities - John Adams, PKU Canada

Emerging PKU enzyme treatment and Australian clinical trials opportunity - Peter J Welburn, Eiger Health Consulting Group

My IEM Experience two special feature stories – Louise Conlisk & Koby Poole

Saturday 22

Mental fitness for resilience and wellbeing - Louise Healy, MDDA Vice President

PKU & The Brain. John Adams, PKU Canada

Neuropsychological assessment: What is it, and what can it offer children and adolescents with a metabolic disorder? - Julia Shekleton, RCH Vic Neuropsychologist

Formula Overview & nutritional considerations for PKU & Other IEM children - Annabel Sweeney, Manager of Nutrition, SA Paediatric Metabolic Clinic.

Formula Overview & nutritional considerations - PKU adults - Emma Clover/Daniela VanderMerwe, Adult Clinical Dietitians, SA

IEM / PKU Research Initiatives, emerging treatments and national care guidelines - Louise Healy, Bianca Albanese & Drago Bratkovic, Unit Head, SA Paediatric Metabolic Clinic.

Emerging PKU treatment and Australian clinical trials opportunity #2 - Neil Smith, Censa

Q&A Panel – Drago, Daniela, Emma, John, Bianca, Julia - facilitated by Louise Healy

Identifying and supporting executive functioning difficulties in children with metabolic disorders. – Julia Shekleton, RCH Vic Neuropsychologist

Interactive Cooking Masterclass - The Grand Hotel Warrandyte

Special Dinner speakers author “*Robin and I explain PKU*” and illustrator PKU child now adult - Jane Brummitt & Caroline Thorpe.

Sunday 23 September

MDDA presented on the current projects underway. We also took this opportunity to gather input from attendees to ensure our projects remain consistent with the strategic goals and objectives of the MDDA and meet the needs of our members. MDDA aims to deliver each of these projects in the first 6 months of 2019, a very busy and exciting time.

PKU/IEM Dietary Management & Wellbeing App – We are so excited to announce the development of “My Low Pro Pal” app has commenced. App feature requests from the retreat have been prioritised and coordination of essential content is well underway.

Volunteer Portal – A volunteer platform has been identified where we can promote project volunteer roles. This will enable us to coordinate resources and tasks with our members and other volunteers to match roles throughout MDDA in which they are interested and wish to provide valuable input.

Transitional Outreach Programs - PKU/IEM Pathways has been scoped providing member support information and resources to parents, families and individuals relevant to each stage in their PKU/IEM transitional journey. Early 2019 will see the first *PeeKabU* patient support bag rolled out. The support bags are designed to provide relevant information and materials to parents of newly diagnosed infants. *PKU Pathway compendium* and resources are being developed for carers' of high needs PKU patients of non or late diagnosed adults.

We would like to thank the MDDA for encouraging us to come to our first retreat. We are leaving with a happy and positive mindset around our PKU challenge and feel so fortunate to have this support.

Ashlee

Kids Club Program

Kids of all ages were well looked after. It didn't take too long for those few who were a little hesitant to join in all the fun. Our focus is on ensuring that all children can experience a range of exciting activities, educational and social that encourage group bonding and develop peer relationships that they can 'take' with them until next they meet. The kids program included:

- Nutritional Educational session & menu planning
- Shopping activity and Low Pro hands on cooking activity with our friends from the Grand Hotel
- Cupcake decorating
- Games Room, video stories and books, mini golf, jumping pillow, fun activities

Weekend Menu Options

The food, low protein options and the venue's attention to detail was outstanding. There was an excellent menu variety across the weekend. We were fortunate that **Ben Van Tiggelen, Executive Chef at The Grand Hotel Warrandyte** designed and prepared onsite the low protein menu, such a treat, thank you Ben! A combination of varying protein free and low protein options were made available for both adults and kids. Protein counts for all lopro options was provided during each meal along with a list of ingredients and sets of scales. Protein for all lopro meals is counted as per ASIEM & MDDA endorsed National PKU guidelines.

The MDDA thank the South Australian metabolic clinics at the Women's and Children's and also Royal Adelaide Hospitals for their support of this year's retreat, and also our many sponsors, in kind partners, speakers and volunteers that all played a key role over the weekend.

Acknowledgements

The MDDA wishes to acknowledge the following partners and supporters of this retreat;

Gold Sponsors



Innovation in Nutrition



B:OMARIN[®]

Silver Sponsor



In Kind Sponsors



EDUCATION & LEARNING



A fantastic weekend with great interactions, topics, and people. Connecting with individuals helps put a face to PKU. It isn't just a genetic disorder, it's people with lives, dreams and ambitions just like everyone else.

Neil

As a grandparent, most of the sessions on the 1st, 2nd days were very informative, enlightening and instructive while I felt I had a reasonable knowledge of PKU, the Retreat has further developed my knowledge and understanding. Thank you.

Lorraine

SHOPPING & COOKING



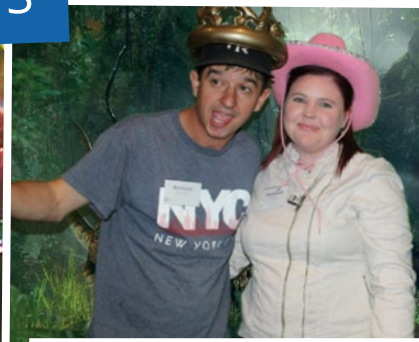
CATCHING UP & CHILLING



After nearly fifty years involvement with PKU we loved being part of this heart warming retreat. Congratulations to the inspirational organisers for bringing us up to date with so many aspects of PKU.. It was lovely to share happiness with so many families. No wonder their children enjoyed it so much that we didn't notice any signs of tears. We adults shared so much, just as children did. Hahndorf was a lovely place to be and catering was brilliant, thanks to the Grand Warrandyte.

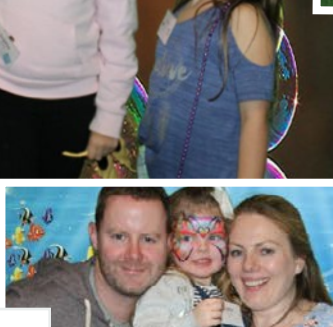
Jane

PHOTO BOOTH SNAPS



I feel honoured and fortunate to be able to be involved with amazing people! Love the PKU/IEM community and making a difference in PKU in peoples lives is amazing, love my friendships, amazing people

Debbie



Fun for all
- extremely informative -
creating PKU family community bonds for life. Reminding us we are not alone and help find solutions to our shared challenges and inspired by innovative research underway.

LeeAnne



My MDDA Retreat Experience

I knew my excitement was building when I found out this years Retreat was in the Adelaide Hills at Hahndorf Leisure Park and Convention Centre because it was so close to home and I found out in time to go! It was also my first time going to a MDDA Family Retreat.

I was asked to speak on the night of the Saturday buffet dinner with Jane Brummitt presenting our journey through creating and publishing our book on people with PKU, "Robin And I Explain PKU". I was so chuffed but a bit nervous about it. I agreed, thinking I know the book well and I'm living it's very story and lessons, so I am pleased to talk about it.

I had never been to the Leisure Park and Convention Centre before and the grounds were very welcoming with lots of grass areas, big gum trees, ponds with ducks, play grounds and wonderful facilities for the families of the event staying in accommodation there.

Families and guests and MDDA staff all milled around chatting then settled in quickly with the first of the talks on the Friday of the 3 day event. I was surprised at how many kids and families came along and those that knew each other caught up and us new comers were welcomed openly.

I was so glad to see other adults with PKU and didn't realise there are quite a few of us. It was amazing to see all the kids. Some with PKU and some without being their siblings. The best part of seeing them all was how happy they were! Unless you knew the families you had no way of telling apart which children had PKU and which did not and to me that is so heart warming because I remember as a child I didn't want to be seen as so different from everyone else because of my diet.

The whole feel of the Retreat was comforting. To know these people all knew about the Metabolic disorder I have and I didn't have to explain it all to dozens of people that I newly met.

Who knew so many people were so close and yet so far with the same condition and their families were so supportive and sharing of their experiences, even from the beginning? It was a special feeling talking with these people.

I found the talks of the next day many and varied and so interesting.

A lot of it comforted me, educated me and confronted me. There were aspects of all the talks for everyone. I also liked that the kids could be free in activities and have fun while we were having our meetings and information sessions.

Another favourite part of the Retreat was the food. The feasts as they were! Tables full of fresh low protein buffet food new recipes and favourites and all so tasty! There was plenty of food for non PKUers as well.

On the Saturday evening we enjoyed a wonderful Restaurant Buffet and I was a bit nervous about my talk but I realised I not only knew the subject well but everyone around me was now familiar and so friendly! I spoke from the heart and was honoured to be asked to speak by the organisers.

I honestly had no idea how much the book, my drawings are in, had reached so many people and still does every year! It is an aspect of my life I am so grateful to have been involved in because the book helps people. Whether it informs, or relieves people or simply makes them smile it has all been worth while.

To be told and see kids enjoying the book is amazing, and adults that grew up with it still love the book. Myself included.

I am truly thankful to the parents that approached me to tell me how much the book means to their families. It is a dream come true to reach people and make a difference in some way through my contribution on such an enjoyable adventure.

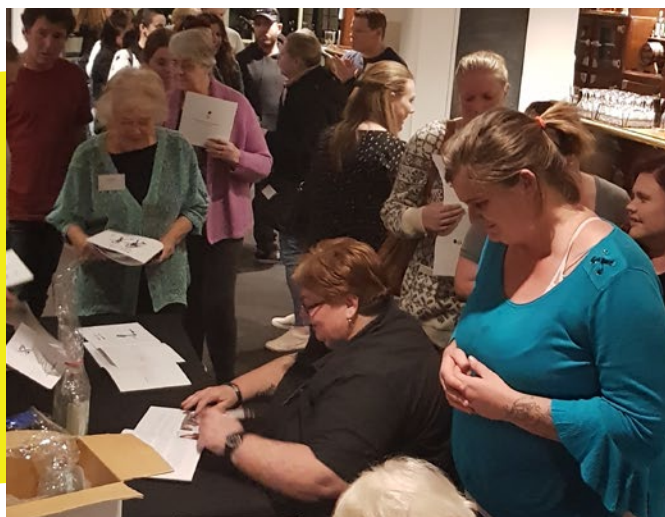
I know everyone involved put so much time and energy into making the Retreat great. It was a fantastic first time experience for me to be with so many people involved and close to PKU. I thank each and every person that arranged and attended the Retreat and made such an important impact on my life concerning PKU. For a long time I had felt very isolated in times of struggle with PKU and alone in empathy about PKU. I no longer feel alone in this and MDDA is an integral part of bringing a lot of us together so none of us ever feel that way again or ever.

Thank you all. Be happy.

Caroline Thorpe



Jane Brummitt & Caroline Thorpe



At the book signing

Connecting with MDDA

Our PKU journey has had positive incremental steps. Firstly, the discovery of another PKU mum through a mutual friend who introduced us up to the larger PKU community. Secondly, joining the MDDA facebook group which saw us being exposed to people's from all experiences including challenges, successes and recipe ideas. However, it wasn't until recently when we attended the MDDA retreat in our hometown of Adelaide that we saw and felt the excitement and enthusiasm from individuals, parents and extended families as well as relief we felt as we saw these grown individuals and children living and thriving with no limitations but a diet, it was then that we saw hope.

The conversations and relationships we formed and the incredible connection we had with people we'd never met before, we form such a strong bond all because of our connection to PKU. I feel that this would not have been possible without the retreat. The information and discussions had with John Adams PKU Canada President, Metabolic Doctor's, Scientists, the MDDA team and PKU individuals leading normal lives this was a weekend we'll never forget and we cannot wait until the next MDDA retreat

We were first time parents trying to figure out what to do with a baby let alone have to learn about PKU. If it's anything we could've changed would be to have joined the MDDA family earlier because the support has been exactly what we needed and our lil man Luca won't grow up alone or different with this condition as he already has extended brothers and sisters who understand because they have a metabolic condition too.

Maria & Ilia

2019 national family retreat

TASMANIA ■ 4-6 OCTOBER 2019 ■ TAMAR VALLEY RESORT, GRINDELWALD



Set on over 150 magnificent acres amid numerous spectacular lakes with breathtaking views. Tamar Valley Resort is located 30 minutes drive north of Launceston airport.

Meet other members of the MDDA community and their families in a fun, social environment.

Educational sessions and workshops
Health Professional presentations
Team building exercises
Kids, Teen club & more!

Registrations will open early 2019. Don't want to miss out?

Register your expression of interest NOW

www.mdda.org.au



A mothers realisation

PKU



Almost 3 years ago we received the phone call from PMH and one of the paediatricians asked us to come as quickly as we could to the hospital as Gabriel's heel prick test was over normal in some parameters, so he needed to be retested.

One more thing she added: "While you are driving to the hospital, please don't Google anything on phenylketonuria".

Of course, curious, journalistic and the anxious part of me Googled PKU straight away and I started crying hysterically. I read scary things and I couldn't believe that this angel like newborn has the rare genetic metabolic condition. I cried so hysterically that my husband nearly lost control over the vehicle. I just screamed, scared my kids to death and deeply wanted to end my life.

So I don't have to take responsibility of looking after him. Some part of me was scared, wanted to escape and hide. Cave.

Over the next couple of months, I lost my warrior and can-do attitude and humour. I nearly ended up with post-natal depression. Some important people pulled me out of that hole I was heading.

Oh god, how deep you can go and end up when life challenges you? Gabriel was just 10 days old.

At Perth Children Hospital we met with a paediatrician, metabolic nurse and dietitian. I remember that dietitian offered some basic human comfort and warmth. She was there. She actually is always there. I remember how I hugged her and just cried on her shoulder, I didn't really want to hear about chemistry, science and genes and how and why and when...

She said that he will do great in life and smiled. At that moment she reminded me of Mother of God. This peaceful and calm moment and I'm sure she had saint's glory shining over her head. These moments are cemented into your memory and even now thinking about this phone call, I turn into tears.

This was the turning point for me.

For some time I thought I was in very dark place imagining that I have been buried and punished, but if I only knew that I was actually planted.

Folks, I know some events and happenings in life are too brutal & savage, it seems like it is too much to handle and carry, but you will need to turn it around in your favour. You get stronger, you revise your values, you grow, become the better person.

Thanks to hard times.

It is part of growth. I'm so glad our life turned this way. This is not the easiest road, but definitely rewarding, challenging and game-changing.

You are turned upside down on molecular level and it's not a bad thing to happen, to be taken out of your comfort zone. I truly believe that we grow through what we go through.

Now three years later, our happy boy Gabriel knows very well what food he is allowed to eat and what food makes him unwell. He loves cooking, he knows how to measure out ingredients for his morning porridge and is very punctual with supplement intake. His biological clock reminds him and he reminds us that is the time for his supplement.

Honestly, his diagnosis changed our life. For better.

Without him we wouldn't be here. I'm very proud of us as a family: eating fresh and healthy, him being on low-protein diet and the rest of family following healthy vegetarian one.

Our son's diagnosis changed our attitudes towards life: your condition doesn't define you and worrying and complaining change nothing. We have never set any limitations on him due to his special diet. We have travelled as a family to various destinations around the world, he loves ballet and we do support his growing interest in dancing.

We never draw any negative attention to his low protein food. So in our house we only say "dairy free cheese" and not "normal and abnormal cheese". We always try his dishes and say how lucky he must be to have specially baked cake, casserole, own pack of vegetable chips.

I believe that positive attitude towards food is very important, not only for kids with a metabolic condition, but for all the little humans who are learning about living and are busy wiring their brains with beliefs, behaviours and sets of attitudes.

The best advice given by one of my friends was:

"Don't play the victim of your life circumstances and don't be guided by fear, pessimism and negativity, be the warrior and be guided by love, courage and positivity."

Viktoria Saar



Dear MDDA family,

Upon reading Trudy's story "Diet for Life" in issue # 67 September 2018, I felt compelled to put pen to paper the moment I read three simple little words...

"especially my mum.."

These words immediately brought tears to my eyes.

My name is Mary. I live in Adelaide and I have PKU. Wow, it sounds like I'm in rehab! (ha! ha!)

Like Trudy, I am a baby of the 60's (only just ...1969), and have met some of the "wiser" PKUers at functions, conferences and retreats.

When we all talk, one thing always rings true. FAMILY plays a major significant role in managing PKU but, hang on,... MUMS of PKU children – they go above and beyond!!!

These super heroes are AMAZING!!

When I read Trudy's story and then think of Debbie Colyer's mum, I can't help but think of my beautiful mum, who was very young, new to this country, and had no idea of how to explain this situation to her husband (who was still learning English at the time).

In my opinion mum's of PKU children all deserve an award.

The mums of us older PKUers understood and persevered with how important it was to be on diet. They were often categorised (by me and probably others) as the "nasty mummy" giving us horrible tasting 'medicine'. I myself was quite a runner and often had my mum, dad and aunt chasing me to give me my Aminogram.

Short note to Trudy: Cynogram must have been disgusting if you thought Aminogram tasted Ok!!!

I have been on diet for life with a short on-off period when travelling overseas in my younger years. My three children are in their teens and not one of them has PKU.

I look at how far PKU developments/educations/screening and food have come over the past 25 years. How lucky children with PKU are nowadays.

A letter from the heart



My beautiful mum, Georgia and my gorgeous daughter Marta

It's almost fashionable to have PKU. You know being vegetarian or Vegan is trending right now. People with PKU are just more disciplined and dedicated.

On a recent cooking day in Adelaide, I met a number of gorgeous babies, toddlers and young children. It truly made me smile seeing how involved both parents were in their children's management of PKU. Raising children these days is a two parent commitment. It was a little different when I was younger. My "ethnic background" resulted in my father constantly working away earning a living for his young family so my mother was home caring for the children. He was a fantastic father but deep down I know that it was mum who had the hardest job of all.... raising a great, healthy PKU child.

Thank you so much to all PKU parents but a special thank you to mums of PKUers. We now understand how difficult it was. I'm sure I speak on behalf of many PKU adults when I say we truly appreciate the sacrifices you made and everything you did for us.

We love you.

Mary Knezevic

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.



END OF YEAR CELEBRATIONS

VIC



Sunday the 18th of November saw MDDA end of year celebrations happening in three states. It is wonderful to see these member events being supported.

It was such a beautiful day for the Victorian catch up with the location being The Grand Hotel Warrandyte. The upstairs balcony provided a lovely spot to chat and sample some nibbles from the low protein menu while overlooking the picturesque Warrandyte river and township. Lunch was a regular and low protein burger and curry buffet. It was great to have a few new families come along, two with new PKU babies – welcome to our community! Great food as always by The Grand and thanks to the Helling's family for organising the event, our Santa and Elf helper and Nutricia for sponsoring.



QLD



Queensland's end of year celebration was held at the Taylor Range Country Club. There was a great turnout with many familiar faces as well as some new PKU families. The lolly hunt and present time with Santa proved to be the day's winner with the kids and adults! Age was no barrier, fun was had by all.

Everyone took a plate of low protein food to share, with there being some lovely new ideas for Christmas treats.

Members in Queensland save the date for 2019 - November 24th. Thanks to Louise Healy and Anita for organising.



NSW



This year's New South Wales IEM party at Parramatta park, had the great weather as well, the sun was shining and the park was abuzz with activity. It was perfect for a relaxed catch up with family and friends. Even though there was only a small number that attended, those that did found the event most enjoyable. Thanks to Debbie and Bianca for organising.

TAS



Thank you to all those who supported the TRC Caulfield Cup fundraising event. Over \$4000 was raised. We really appreciate it! 100% of all funds raised go directly to supporting families with PKU. MDDA are most grateful to the Penneyston Family for their passionate enthusiasm and support of raising awareness and funds to support all families living

ideas

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!

QLD

Fun run

Congratulations and thank you to Bianca Albanese, who on the 16th September, completed the Blackmores 10km Bridge Run. Bianca chose to donate all funds raised to the MDDA. Over \$1400 was raised.



A special thank you to the following volunteers

*Laurel Tulloch & Emily Large
for our knitted IEM baby bears*

*Mel and Darcy, who without them we would
not have got our last newsletter sent out.*

all our state end of year celebration event organisers

*Sasha & Steve Cranwell for storage
and pickup of retreat products*

We really appreciate all your time and effort in supporting the MDDA.

LOPROFIN® PRODUCT RANGE

A low-protein diet shouldn't mean less choice or enjoyment. The Loprofin® range offers quality low protein foods that help increase variety in your diet.

- Purchase online through **PKU Connect**
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BREADS



SNACKS



PASTA



RICE



FLOUR MIX



CEREALS



MILK
SUBSTITUTES



EGG WHITE
REPLACEMENT



EDITORIAL

In late 2017 the PKU Association of NSW Inc. were honoured to receive the prestigious “Founder” award from the Children’s Hospital at Westmead for donating in excess of **\$1 Million dollars to PKU research**. This is such a fantastic and amazing achievement considering the number families that support our wonderful association. We could not have funded the valuable streams of PKU research if it wasn’t for our amazing, supportive families, friends and donors.

As the year is almost coming to an end the PKU Association of NSW Inc would like to provide an update of where we are with the research activities.

The Children’s Hospital at Westmead

“Stop the Nonsense” - Potential therapeutic for PKU Dr Minal Menezes

About 10% of individual with PKU have a so-called nonsense PKU gene mutation, where the mistake causes the PKU enzyme to prematurely stop being made beyond the point of the mistake in the gene. This results in a shortened PKU enzyme called phenylalanine hydroxylase deficiency (PAH), that is either unstable and falls apart altogether, or which is otherwise non-functional. Such nonsense mutations are often associated with more severe PKU, where dietary phenylalanine tolerance is markedly reduced.

A potential therapy for PKU is the ability to restore a functional PAH enzyme. Research in other genetic disorders has shown that there are a number of drugs, which can trick the cell machinery to “read through” the premature stop, allowing a full length enzyme to be made. A major problem at present though is that most of these drugs that can be used as “read through” therapy are toxic, particularly affecting kidney function, and so cannot be used to treat patients. However, we are aware of a new safe “read through” drug, which has been clinically approved for many years and has been used for its function as an anti-inflammatory antiallergic agent. Once we have successfully established the efficacy of the drug in a cell based system, the next step would be a pre-clinical trial. This will demonstrate the translational capacity of our research in identifying an effective and safe therapeutic treatment for PKU patients.

Anti-aggregation agent is a potential therapeutic for treatment for PKU | Dr Minal Menezes. The second area of our research focuses on a subset of missense mutations of the PAH gene are known to cause aggregation of the PAH protein i.e. makes the protein sticky. This is hypothesised to be a result of misfolding of the mutant polypeptide. Arginine is a commonly used safe supplement but surprisingly also has protein stabilizing effects. Preliminary cell based studies with arginine have shown anti-aggregation effect, leading us to hypothesize that it might constitute a promising therapeutic agent in specific cases of PKU. Further investigations are in progress to assess the effect of arginine on PAH protein levels and function.



Adult MRI Studies | Dr Michel Tchan

This project aims to perform advanced neuroimaging studies on patients with phenylketonuria (PKU), with a view to determining the role of disease management on subtle measurements of brain structure and function. This project will perform advanced brain MRI scans on 90 patients with PKU, and simultaneously assess their neurocognitive function. The information obtained from these studies will provide further knowledge about the long term impacts of PKU on brain structure and function.

The Children’s Hospital at Westmead and The Murdoch Institute

Development of a Genetically Modified Probiotic for treatment of PKU | Prof. John Christodoulou

The team are continuing to generate and evaluate the effectiveness of a genetically engineered probiotic species, which contain a phenylalanine ammonia-lyase (PAL) gene of plant origin, and which will have high-level expression of the PAL protein in vitro and in vivo. By presenting the gut with genetically engineered probiotic, there will be high levels of functional PAL enzyme in the lumen of the intestine, which will decrease the load of phenylalanine available for absorption across the gut, and consequently would result in a reduction of toxic blood levels of phenylalanine.

Bridie Melham



PKU Youth Camp

The PKU Youth camp at Berry in NSW was held 2-6 October. Congratulations to PKUNSW for another successful camp. Some activities the happy campers enjoyed were cooking, rock climbing, dodge ball, and canoeing. They were also lucky enough to have a visit from the staff at Westmead Children’s Hospital, who helped educate on protein counting and PHE levels.



Recipes

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



Christmas Biscuits

Ingredients:

50g Butter
50g Margarine
175g Low Protein Baking Mix
1 teaspoon ground mixed spice
3-4 teaspoons cold water

Method:

Preheat oven to 150°C.

Beat the butter, margarine and sugar in a large bowl until turns pale and fluffy.

Add the Low Protein Baking Mix and spice, stir into the creamed mixture.

Squeeze the mixture together until a dough starts to form, if necessary add a little water to help bind the dough.

Transfer dough to a surface lightly dusted with the baking mix. Knead for about 30 seconds until smooth.

Roll out the dough to 1/2 cm thickness and using a Christmas cutters cut into desired shapes.

Place the biscuits on a tray lined with baking paper and cook for 20-25 minutes until firm and slightly browned.



Cinnamon, Cherry & Pear Muffins

Ingredients:

1 cup Low Protein Baking Mix
2 teaspoon Baking powder
2 tablespoon Castor sugar
½ teaspoon Cinnamon
1 teaspoon Egg white replacer
50g Butter melted
½ cup Low Protein Milk substitute
½ cup canned seedless cherries drained and halved
½ cup canned pears drained and chopped
Icing sugar to serve

Method:

Preheat oven to 180°C and lightly grease a 4 muffin pan. Sift Low Protein Baking Mix and baking powder into a medium mixing bowl.

Add castor sugar, cinnamon and egg white replacer, stir to combine.

Combine milk substitute and melted butter, make a well in the centre of flour, and add milk mixture and fruit. Stir with a metal spoon until just combined.

¾ fill each muffin hole with mixture (approximately ½ cup) and bake in the oven for 25 minutes, or until a skewer comes out clean.

Let the muffins stand in the pan for 5 minutes before turning out onto a wire rack to cool.

Serve dusted with icing sugar.

Root Vegetable Crumble

Ingredients:

250g potatoes
250g carrots
250g parsnip
3 tablespoons olive oil
2 tablespoons mustard powder
3 tablespoons apple juice
Salt and pepper to taste
180g low protein baking mix
60g crushed low protein cereal
125g Nettlelex

Method:

Peel and wash all vegetables and chop into bite sized chunks.

Plunge them into boiling water for 6 minutes (no longer).

Reserve 4 tablespoons of the cooking water and drain the rest.

Put olive oil, mustard powder and apple juice in a basin, add the cooking water and stir, this becomes a sauce.

Arrange the blanched vegetables in a shallow baking dish and pour the sauce mixture over. Toss until well covered. Season with salt and pepper to taste.

Combine the low protein baking mix and crushed low protein cereal in a bowl and rub in the Nettlelex.

Sprinkle the crumb mixture to cover the whole surface of the vegetables in a baking dish. Dot with extra Nettlelex and bake in a moderate oven 170°C for 40 minutes.

Serve hot and enjoy.



Product update



Woolworths free from gluten:

Fruit mince bites

Protein per 100g: 5.3g
Protein per serve: <1g
Serving size: 6g
Available from:
Woolworths



Leda: Dunk-ies

Protein per 100g: 4.7g
Protein per serve: 1g
Serving size: 22.5g
Available from:
Coles & Woolworths



Simply Wise: Custard Pastries

Protein per 100g: 1.8g
Protein per serve: 0.56g
Serving size 30g
Available from:
Coles & Woolworths



Healthy Heart: Cauliflower rice

Protein per 100g: 1.5g
Protein per serve: 2.25g
Serving size: 150g
Available from:
Woolworths/IGA



Fruit for Life: Crazy Coconut- coconut chips

Protein per 100g: 4.3g
Protein per serve: 0.9g
Serving size: 20g
Available from: Woolworths



The Wise Bunny: Roasted Almond Milk Yoghurt - Mango

Protein per 100g: 1.6g
Protein per serve: 2.4g
Serving size: 150g
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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