



MDDA NEWS

Newsletter of Metabolic Dietary Disorders Association

Issue 57
2015

Happy Easter!

Our office will close on Good Friday 3rd April and reopen on Tuesday 7th April 2015.



Looking forward 2015

The start of the year has been a busy couple of months for the MDDA. With submissions and grants all due and the WA Retreat fast approaching it has proved to be a very productive start to 2015.

The **WA Retreat** is only weeks away. If you can't make it don't despair our next retreat is headed for Queensland 21-23 August at the Alexandra Park Conference Centre, Alexandra Headland.

Last year we welcomed on board **38 new members**. We look forward to continually spreading our resources and knowledge to others and always welcome new members and new ideas. Our facebook support group has grown along with our members fundraising efforts.

In this issue we share with you some inspiring first day at school stories from four of our younger members. Fun **Easter recipes** for you and your children to enjoy along with our usual product updates.

Included is information on our lobbying plans and details on the **National Clinic Roadshow** held over the closing months of 2014. You will find an update from Prof John Christodoulou making steady gains on the PKU Research front.

We have many events and activities planned for 2015 and look forward to your involvement. Make sure you keep familiar with our website www.mdda.org.au. We are regularly updating information, upcoming events, pictures and articles. We are also close to launching our **recipe data base**. Soon you will be able to search and with the press of a button have a range of recipe ideas at your fingertips. A new **product catalogue** will also be launched. We will be requesting that you all contribute to make these resources a valuable tool to assist with your management of an IEM.

We look forward to the oncoming year and hope to see you at one of our events.

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**Donations over \$2.00
are Tax Deductable**

Transitioning to Primary School

Charlie Cooper (age 5)

Prep - North Ringwood Primary School, Vic

How was Charlie's first day of school?

Very exciting! Charlie has been waiting to go to school for soooo long – so he was just full of beans on his first day of school.

How was Charlie in the morning/ how were you?:

Up early and ready to go. No nerves just excitement and chuffed to be doing what his big sister does. I was excited too, and just fed off his energy so there were smiles all round.

What did the school/teacher know about PKU?

Nothing until I wrote a letter to the school and then met with the principal and teacher to discuss Charlie's condition and needs.

How did the teacher/school react to any special requirements?

Very positively. To the extent the principal himself has even gone out and sourced his special 'bubble and squeak' patties to have for him at school BBQ's.

Are there any major difficulties you have experienced with your child starting school?

Just keeping him fed with healthy snacks that also fill him up but keep his levels down. Charlie's appetite has almost doubled over the summer so I am needing to stack his lunchbox full of 'heaps' of food, and then including an emergency snack in his bag pocket so once he has finished his lunchbox he has something if he needs it. When he gets hungry he goes into 'meltdown'!

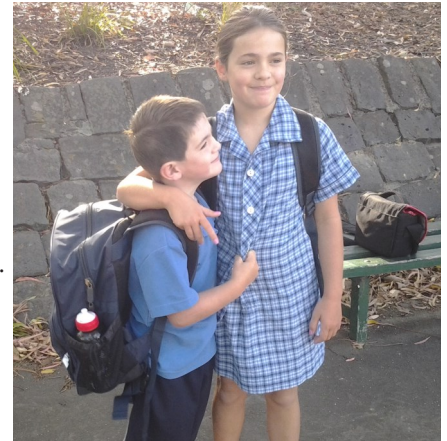
What is generally in Charlie's lunch box?

A honey sandwich on white bread, or country life wholemeal, a low pro banana bread or carrot cake (homemade), an LCM bar, a fruit squeeze, a fruit pole, a banana, a bag of grapes, low pro popcorn or rice-cakes, a few slices of bio cheese and lo-pro crackers = generally around 6-7 grams (as he is permitted 12 in a day so this permits 2 grams at brekky and 3-4 grams for dinner).

Yep I know - he is an eating machine!!

What is Charlie's favourite food?

He is a very versatile and non-fussy eater (yes we are very lucky and I tell you if it was my elder daughter it would be a very different story). He loves honey sandwiches and pku vege/sausages, as well as of course anything chocolate.



How will you deal with class parties?

We have provided the teacher with a special box of snacks he can have in place of anything brought in from another child or parent. This seems to be working well, and the teacher also knows there are some things he can have.

What is Charlie's favourite part of school/Why?

He loves his teacher Miss Easy and of course playing with his friends at playtime. He also loves riding his bike or scooter to school. He also has his first canteen visit the other day too and was very excited that he could buy a frozen juice box!

Any tips for future Prep parents in making the transition to school easy?

Make early contact with the school to ensure they assign a competent and 'nurturing' teacher to your child. Be confident to 'make enough of a fuss' about your child's condition so that they are aware and attentive early. Focus them on helping to promote your child's independence and confidence with their food and formula intake from the very beginning, and encourage a regular positive and open dialogue with your teacher by being present (when you can) at school dropoff or pickup times or during occasional classroom help opportunities.

I also recommend visiting the canteen and going through the list of what might be suitable for your child so that they can occasionally pay a visit to the canteen - like most other kids.

Fundraising Raffle and Entertainment Books

We would like to thank **Tracey Scott** for her tremendous efforts in organising a work raffle and coordinating sale of Entertainment Books in support of PKU research.

The funds raised by Tracey will be donated towards PKU research by Professor John Christodoulou and his team at The Children's Hospital, Westmead.

Tracey is co-ordinating the sale of the Entertainment Books for 2015. Get in early to make use of the full year of offers. Contact office@mdda.org.au or freecall:1800 288 460.



Transitioning to Primary School cont

Sophia Haines (age 5)

Prep - St Mary of the Cross, Point Cook. Vic

How was Sophia's first day of school?

Sophia was so excited to start school. She had about 3 prep transition days late last year and had already made a few buddies. Her first day of school was all smiles and fun.

How was Sophia in the morning/ how were you?

We were completely fine. Although, we got the keys locked in the car and just made it on time.

What did the school/teacher know about PKU?

The week before Sophia began school I did a presentation to the entire staff about PKU. I showed the Kevin Alexander youtube film. I also had given them a copy of the PKU handbook and copies of the childrens book.

How did the teacher/school react to any special requirements?

Everyone at the school is very accommodating. We have a set time for Sophia to go and drink her formula daily. She goes to the School receptionist each day to have her formula and a communications book is signed off.

Are there any major difficulties you have experienced with Sophia starting school?

None at all.

What is generally in Sophia's lunch box?

We pack a honey sandwich made from Country Life Gluten Free Bread (2.4 grams). Or a salad wrap made with gluten free wrap (2 grams). A portion of fruit (Grapes or Strawberries), sometimes olives or sultanas. Lo-pro crackers and carrot sticks.

What is Sophia's favourite food?

Wrap

How will you deal with class parties?

The teachers asked me to provide them with treats at the start of the year so I gave them a big bag of all kinds of lollies and kids party bags. All the kids are given treat bags



to take home. They do not have cakes or cup cakes so it is quite easy.

What is Sophia's favourite part of school/Why?

Recess, Sophia loves the monkey bars.

Any tips for future Prep parents in making the transition to school easy?

Communication, I would say. We chat to the teachers every day and the principal. There has already been a pancake day which we were made aware of well in advance so we could make lo-pro pancakes for Soph.

Maggie Marshall Fundraising Thankyou

In October 2014 Western Australian members were lucky enough to be able to attend a quiz night, organised by Scott & Fiona Greer, Maria & David Contera and Vanessa Mekisic, to help raise money and awareness for the MDDA.

Maggie Marshall, a colleague of David's at St John of God Healthcare Murdoch, was unable to attend this event and organised instead a raffle to raise funds on behalf of the MDDA. It was a great effort raising \$210. We are so grateful to Maggie for her wonderful efforts and support.



Martin Buckland, Maggie Marshall & David Contera

Transitioning to Primary School cont

Cayden Mitter (age 4)

Prep - Clover Hill Primary School, Qld

How was Cayden's first day of school?

Cayden's Kindergarten spent a lot of time preparing the Kindy kids so they understood what a bell would mean, lining up for class and they structured their classes very similarly to a prep day so Cayden was very well prepared for what the day would be like. He was excited to wear a new uniform and see his friends from kindergarten who he hadn't seen for a few weeks. It might have felt like more of a social thing for him than a "school" thing.

How was Cayden in the morning/ how were you?

Cayden was proud and excited to be going to big school where a few of his other friends already had been attending for a year. The morning was fine, when the bell went the teacher (who Cayden had already met) welcomed them in and he said goodbye and walked in, no tears. I, on the other hand, felt a lot of nerves and tension. I was afraid for him and wondered if the formula was going to be a problem. I don't worry as much about Cayden eating others food as he is very good like that (so far).

What did the school/teacher know about PKU?

The school met with me first after I emailed them to ask how they would manage Cayden's PKU requirements. Interestingly I emailed three schools in the local area (that we could have attended) and Clover Hill was the only school that responded to my email and their response was on that same day. I felt we were off to a good start with that. I sat down with the enrolling officer who took as much information as she could from me to mark in Cayden's file. She talked me through the tuckshop system and what food based activities the school runs (i.e. sausage sizzles etc.).

There was one admin teacher at the school whose wife was a nurse so they had heard a little about metabolic conditions however until I brought in the information and sent through links for the teacher to read both the school and Cayden's teacher had never heard about PKU.

How did the teacher/school react to any special requirements?

Very, very well I must say. I have been very lucky with both Kindergarten and school being keen to help and alter the classroom environment in anyway possible so Cayden can partake. Our teacher asked me what I would like to see happen with regards to Birthdays. Her attitude is that if one child cannot partake in an activity then no child shall and so she wanted to know if I would like the children to not bring in Birthday cakes to share with the class. I asked if she would be happy for me to send a cupcake to be stored in the freezer as school and de-frosted when there is a party and she was more than happy to facilitate this for us.

The tuckshop so far have held sushi days, pirate days and frozen days at school where there are special things on the menu that can be ordered. For sushi day I made my own sushi and sent it to the tuckshop for them to delivery to Cayden with the other children, on Frozen day he was able to eat the apple sorbet that they made (I checked

all ingredients with them) and the pirate day that's coming next week they have potato gems and carrot swords with avocado dip so Cayden is able to partake in that day also. The tuckshop have asked me to come to them with any requests or queries and they will do all they can to accommodate Caydens needs. They have his face on the board and know that he is not able to purchase anything outside of the items that I have checked with them.



Are there any major difficulties you have experienced with Cayden starting school?

Cayden is still refusing to take his formula out of anything other than a bottle. His teacher has made allowances for this and informed the class that this is the best possible method for him to consume his very important medicine. Some of the children in Cayden's class were from his kindy so they all already knew about Cayden's formula and I believe that has helped him. We tried again the other day to get him to have a taste of a cooler and he vomited after one sip. That's not particularly school related but it is a challenge.

What is generally in Cayden lunch box?

Cayden's school has a healthy snack time at 9:30am so he will eat an apple or pear, and a fruit pole or dried fruit for this break. Morning tea is at 11am and he will eat a jelly, bio cheese and crackers, or a packet of potato sticks and lunch he will eat a sandwich (white bread with crusts removed with promite, honey or jam) and a piece of cake - home made slice or LCM. Occasionally we will send a wrap with cream cheese (Tofutti brand), avocado, grated carrot and lettuce (small amounts as he is picky) but I don't think this fills him up as much as a sandwich tends to.

(Cayden has an allowance of 10.5 grams of protein per day and the school lunch box can be anywhere from 4 - 6 grams of this allowance making breakfast always low protein and dinner and dessert low also but at least his lunch box looks similar to others)

What is Cayden favourite food?

Cayden loves his pasta and cheese sauce with vegies. He has a sweet tooth and thinks a bowl of ice cream (homebrand) at the end of the day is almost too good to be true so I make him work for it.

How will you deal with class parties?

We try to find out what is being supplied at the party and make something as similar as possible. If Cayden gets a slice of fairy bread he is chuffed with that. He isn't big on

(Continued on page 5)

(Continued from page 4)

potato chips or corn chips so we stay away from that anyway. I will take some marshmallows and take a toy to swap for any chocolates that might be in a party bag.

What is Cayden's favourite part of school/Why?

I ask each day what he did today and he can't ever seem to remember..... So that has started already. He does love his teacher and says she is the best teacher he has ever had (I think he said that last year also about his Kindy teachers).

Mrs Walker runs a program in her class based on doing nice things for one another. If she sees that you have been a nice child (be it saying nice things or helping one another) she puts a pompom in your bucket and you have been "A Bucket filler" for the day. If you have been

extra good you might get a bracelet to say how extra good you were at filling your bucket. He seems to love being recognised for his efforts. He enjoys Science class from what I can gather and seems fascinated by clouds ever since they learnt that clouds contain rain. I love watching him learn about things that we seem to just take for granted.

Any tips for future Prep parents in making the transition to school easy?

Have as many conversations that you can with your teacher and school. Build a relationship with your teacher and the teachers aid in the classroom, I email Cayden's teacher almost weekly to let her know how his blood tests are tracking and if she has noticed anything. It has worked a treat for us so far.

QLD FAMILY RETREAT 2015



**21-23 August 2015
Alexandra Park Conference Centre
Alexandra Headland, Sunshine Coast, QLD**

This retreat will provide an invaluable opportunity for people with inborn errors of metabolism and their families to meet in a social environment and build positive relationships with other members of the MDDA community.

The proposed outline will have emphasis on community support, interaction and sharing. The focus is for a relaxed, informal environment where you will make new friends, learn new ways of coping, learn new food ideas and leave with a sense of wellbeing. We would like to offer a range of sessions and breakout groups to build a foundation for your new skills. We invite you to submit ideas of what you would like to see included.

These events will be heavily subsidised by the MDDA. Please register your interest with the National Office at office@mdda.org.au or 1800 288 460

Give Happy Live Happy National Volunteer Week 11 -17 May 2015

MDDA value all our volunteers whether it's one hour of your time or an ongoing commitment. Thank you for sharing your skills and interests to assist all living with an inborn error of metabolism.

If you would like more information on how you can assist, contact office@mdda.org.au or Freecall 1800 288 460



Transitioning to Secondary School

Brooke Hellings (age 12)

Warrandyte Secondary School, Vic

How was your first day of school?

I really enjoyed meeting new friends and getting to know more about people. At first I found it really tricky to find my way around but now I am used to it.

How were you in the morning?

I was really excited and I met at the bus stop to catch the bus with my friend. I felt a little confused at the start because I didn't know where to go sometimes.

What did the school/teachers know about PKU?

Well Nat and I were the first people to go to Warrandyte High School with PKU. In food technology Nat and I always cook together and we bring our food in. Sometimes they will have the food we need or we bring it to them and they keep it.

How did the teachers/school react to any special requirements?

The food tech teacher gave us the menu so we can discuss what we can and can't have.

Are there any major difficulties you have experienced with you starting secondary school?

Sometimes the food tech is hard because it doesn't always work like the others.

What is generally in your lunch box?

Wrap, bar, popcorn, jumpies, cupcake, fruit, sometimes a hash brown.

What is your favourite food?

In general I like sweets and hot chips but in my lunch box I

enjoy jumpies, popcorn, hash brown & cupcakes.

How will you deal with class parties?

Well sometimes I just eat the food I have otherwise I just pack extra lunch in case I can't have anything.

What is your favourite part of school/Why?

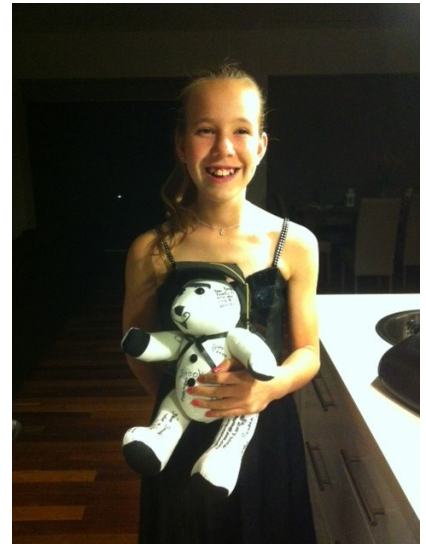
My favourite part of school is P.E, food tech, recess and Lunch. With P.E you can get active and have lots of fun, with food tech you get to create new foods and we can still cook, and recess and lunch because you just hang out with your friends and I really enjoy that.

What is the difference between Secondary and Primary?

I use to leave my formula/coder at the office at primary school, now I leave it in my bag in my locker and have it at lunch time.

Any tips for future Secondary School students in making the transition to school easy?

When you do food tech ask for the menus and when you have it you just give the food you bring and then you can cook. Also when I went to orientation day my school did a sausage sizzle so maybe ask before you go to school if they are cooking anything otherwise bring your favourite easy cooked meal and you can have that.



Last Chance WA Family Retreat 2015



Friday 17th to Sunday 19th April, 2015

Point Walter Recreation and Conference Centre, Bicton, Western Australia

Overlooking the Swan River the Conference Centre is 20 minutes from Perth CBD. The venue will allow for a weekend of family adventure with plenty of activities and fun to be had onsite as well as informative presentations and discussion sessions.

MDDA retreats are designed to be relaxing weekends away to enjoy the company of other families and members of the MDDA, to make new friends, learn new ways of coping, learn new food ideas and leave with a sense of wellbeing and empowerment. They are also highly educational and provide all attendees with direct access to selected IEM healthcare professionals and other specialist presenters.

For further information and registration refer to our **website** or call our office on **1800 288 460**

Meringue Nests

Ingredients

75 ml egg white replacer (as per package instructions)
75 ml water
150g caster sugar
Food colouring (optional)



Method

1. Whip liquid until soft peaks form then gradually add the sugar until the mixture is stiff.
2. Add food colouring if desired.
3. Draw 8cm circles onto non stick baking paper.
4. Fit piping bag with star nozzle, fill with mixture. Pipe a continuous coil of meringue to fill the circle which will make the base, then pipe a ring of meringue on top of the outer edge.
5. Sprinkle with 100s and 1000s if desired and bake very slowly in a low oven (approximately 160°C*) for about 2 hours or until they have dried out.
Before serving, fill meringues with low

protein cream and top with your choice of fruit, low protein chocolate or decoration.

N.B. Make the egg white replacer solution the day before and refrigerate. The mixture beats up more easily if the solution is very cold. Meringues are less successful if made on a humid day.

*Preheat oven at 180°C and when ready to bake the meringues, turn down the oven to between 140°C and 160°C. Bake in a very slow oven and after required cooking time, turn off the oven and leave the meringues to cool completely.

Easter Bunny Salad

Ingredients

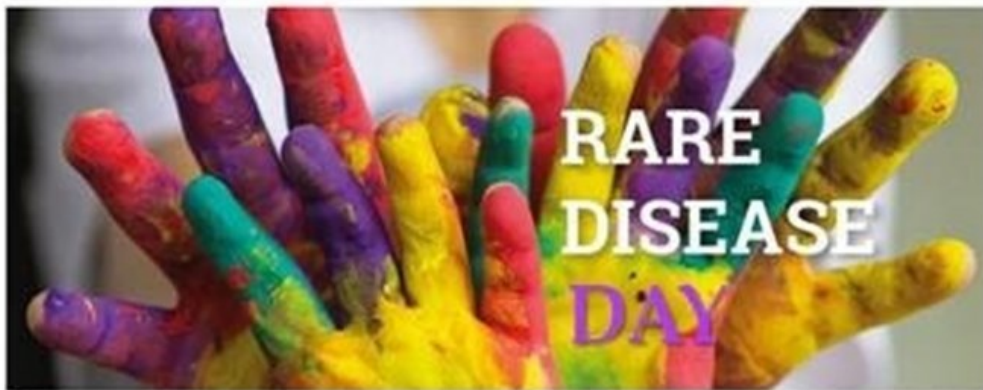
Pear halves
Raisins
Miniature meringue
Lettuce
Apple Slices or other fruit/vegetables as desired

Method

1. Place a lettuce leaf on a plate
2. Add the pear half onto the lettuce for the body
3. Insert raisins for the eyes and nose
4. The ears are made from apple slices and a miniature meringue becomes the bushy tail.

Try some different versions. Decorate and become creative with your Easter bunny. Please send us in some photos.





Rare Disease Day 2015

In recognition of Rare Disease Day 2015 the MDDA attended a presentation hosted by the Genetic Support Network of Victoria (GSNV) in conjunction with Murdoch Children's Research Institute at the Royal Children's Hospital in Melbourne.

The morning began with insights from health professionals and researchers, a parent sharing her personal experiences of having a child with an undiagnosed rare disease and the impact that has

on their family, followed by a panel discussion with the opportunity for questions.

MDDA representatives will also be attending the Rare Voices Summit in March to identify ways that patient support organisations can work together to give a stronger voice to people living with rare diseases.

National PKU Awareness day MAY 1st 2015

We encourage YOU to raise awareness in your local community

Volunteer to host a fun run/walk in your area on Sunday May 3rd. MDDA hopes to run one activity in each state on this date this year and will provide you with plenty of support.

Other ways to get involved

- Ask your childcare, kinder, school, university or work place if you can put up a display on their noticeboard
- Ask your local library/gym, if you can put up a display
- Call your local/national paper on TV and ask if they would like to write an article about a fantastic subject— You and what it is to live with an IEM.



Congratulations to Jill Colyer

Awarded the Order of Australia Medal.

Awarded For "service to community health through advocacy roles. Mrs Colyer has served as an advocate of people born with the rare genetic metabolic disorder, Phenylketonuria (PKU) since the 1960s, seeking advice from dietitians and food associations in the United States of America and promoting a suitable Low Phenylalanine diet for children with PKU in Australia."

Update on Government Lobbying Plans

At the Sydney retreat there were a number of group discussions about the need to lobby government bodies on issues related to living with an IEM and the resources and support available. These issues ranged from equitable access to metabolic clinics, to management of the IEM grant and access to medications via the Pharmaceutical Benefits Scheme (PBS). The consensus from these discussions was that a co-ordinated approach, and some expert advice, was the best way to ensure our voice is heard by the right people. A joint team between the MDDA Executive Committee and the PKU NSW Kuvan team has been formed to consider our strategy

Since that meeting we have undertaken extensive research and sought advice from professional and experienced sources about the best way to achieve a result from lobbying efforts. Based on research and advice the proposed strategy includes:

1. Seeking agreement from government for a national framework for the management of rare diseases in Australia that aligns with countries with similar resources, health systems and disease prevalence – for example Canada. Rare Voices is currently working on this and we will become a part of this campaign.
2. Collect data regarding the prevalence of the relevant IEM's in Australia and current treatment protocols and provide comparison data for how they are managed in similar international communities. The collection of data was discussed with each clinic during our recent clinic tour and we are currently writing to formally request this data.
3. Seek agreement from the Australasian Society for Inborn Errors of Metabolism (ASIM) for national guidelines for the management of IEM's in Australia covering such issues as clinic access and resources, treatment protocols, drug therapies etc. Strengthening the link with our clinics via our clinic tours has given us the opportunity to seek clinical input for this and the National Treatment Guidelines initiative is an important part of this. ASIM's first working party looking at adult guidelines have made significant progress in collating evidence for the first set of guidelines.

4. Based on the results from the three steps outlined above we will design a letter writing and MP visit campaign that is co-ordinated but not uniform. We are advised that personal stories provided in a co-ordinated way are most powerful. It is also essential that the campaign is based on facts and supported by medical professionals with experience in managing IEMs.

This means over the coming months we will be looking for people who want to get involved in this lobbying project so if you are interested and think you could help please let us know.

Lobbying for Kuvan

Based on the research we have done on Kuvan the lobbying for this is a bit more complicated and likely to take more time. We have collected the following information:

- Kuvan is registered by the Therapeutic Goods Administration (TGA) for treatment of PKU. Which means the individuals or individual prescribers can legally use it to treat PKU in Australia if they can pay for it. This means some hospitals may pay for it for some patients where they are tested to be responsive to it (as not everyone with PKU is).
- In July 2011 the Pharmaceutical Benefits Advisory Committee (PBAC) rejected an application to have Kuvan listed on the PBS for the treatment of PKU based on cost effectiveness principles. (You can read about the submission to the PBAC here <http://www.pbs.gov.au/info/industry/listing/elements/pbac-meetings/psd/2012-07/sapropterin> and the finding of the PBAC here <http://www.pbs.gov.au/info/industry/listing/elements/pbac-meetings/pbac-outcomes/2012-07/subsequent-decisions>) This was because taking KUVAN does not reduce the cost of any other treatment such as formula already subsidised by PBS. In that application it was approved for use in patients who have BH4 deficiency, which cannot be treated by diet alone. HPA caused by BH4 deficiency is a much rarer condition than PKU affecting 1-2 babies per 1 million live births. People who have Hyperphenylalaninaemia that is a milder form of PKU (caused by PAH deficiency) are not subsidised for KUVAN.

BH4 deficiency is routinely screened for in the newborn screening process.

- We contacted the pharmaceutical company who distributes Kuvan in Australia and based on the ruling of the PBAC in 2011 they have no plans to resubmit an application to the PBAC. Because making a submission to the PBAC costs pharmaceutical companies a considerable amount of money, they would need to feel confident there is a reasonable chance of success for a second submission. This means that we would first need to convince the company to resubmit before lobbying the PBAC as the PBAC will not have any submission to link our lobbying to.
- We have sought advice from medical professionals and peak bodies with experience in health/medication lobbying regarding the situations under which the pharmaceutical company would resubmit and although not definitive the recommendation is that some sort of government lobbying under a government and professional body endorsed guideline would give us the best chance of the drug company resubmitting. If that happened then submissions to the PBAC could be considered in their decision-making.

Given all of this, our broad strategy is to keep working with Rare Voices and ASIM to get consistent treatment guidelines for both the treatment of IEMs in Australia and also the management and funding of rare diseases such as IEMs in Australia. We could then use that as a platform for lobbying for KUVAN.

Our conclusion is that the campaign to have Kuvan subsidised on the PBS scheme will be challenging and require us to work together as a community. If you know anyone with professional skills and experience in this area who would be willing to advise MDDA on a pro bono basis please let us know.

Louise Healy
Wellbeing Co-ordinator

Visit to Menarini Australia

Our trip to Sydney in December, to meet with the NSW adult and paediatric clinics, provided the perfect opportunity to share with the staff of Menarini the enormous impact their support had made on the Tyrosinaemia families who attended the Sydney retreat in September last year.

A few retreat photos, some feedback from those who attended and a short video from a Tyrosinaemia family were all it took for the staff at Menarini to be visibly moved.

They loved hearing the stories and feedback from those who attended and could really feel at a personal level what a difference they had made with their contribution.

They then confirmed that they will be providing further support in 2015 to the MDDA to help us support HT1 families better.

I found our visit really moving and inspiring, it was obvious from the response how much Tracey and the team at Menarini care about making a difference to their customers. We are really grateful for their support.

Monique Cooper



MDDA State Clinic Roadshow – 2014-2015 Review

Over the past three months MDDA President Monique Cooper and Wellbeing Director Louise Healy have undertaken a whirlwind tour of the Metabolic Clinics around Australia. The tour had a number of purposes:

1. As part of our mission to ensure that members have access to information and resources to enable them to manage their conditions effectively we wanted to strengthen our relationships with the clinics to understand how we can work with them to provide the best support possible to members.
2. Promote the Wellbeing programs we are in the process of developing and launching to gain the support and input of the clinics. These programs are designed to provide tools and support that enable people living with an IEM to help them experience wellbeing in all aspects of their lives.
3. Get a better picture of the number and management of IEM's across Australia so as an organisation we can understand gaps in support for people who are living with IEM.
4. Provide clinics with our new brochure and highlight the free first year subscription to encourage people to become members.
5. Discuss the proposal we submitted to ASiEM requesting consistent National Treatment Guidelines so that clinicians can understand our drivers and we can understand their

position in relation to the guidelines they currently use.

The tour has been a great success, mainly due to the generosity and commitment of the metabolic health professionals around the country. We are a very lucky community- our doctors, nurses, dieticians and social workers are passionate and very knowledgeable. Their commitment gave us plenty to talk about and Monique and I came away with a new understanding of the breadth and complexity of metabolic dietary disorders and a new appreciation for the broad knowledge and skills our medical professionals have.

As a result of this tour the MDDA Executive team will be meeting to review our priorities and strategies for the next 2 -5 years. The tour highlighted important issues for our community that we need to give focus and attention to. Key priorities we have identified include:

- Government lobbying for medications and resources.
- Increased public awareness of metabolic disorders.
- Fundraising to support a broader range of research projects including metabolic disorders and aging, health impacts of diet, transitions and best practice models for metabolic care.
- Ongoing dialogue with clinicians to ensure support services we provide meet community needs and that all

families who are managing an IEM have an opportunity to access support if they want it.

- Maintaining strong links with related organisations such as Rare Voices and the Newborn Screening working party to ensure that we influence policy and decision making in these areas.
- Continuing to provide opportunities for members to connect at events and retreats.

We will be announcing more about our plans as they evolve and there will be many opportunities for members to get involved in projects. We would love it if you could think about areas where you would like to contribute and then watch this space as we ask for involvement on a range of projects to achieve these priorities.

Thanks to Jenny for her practical support in helping us to organise the tour, to all the members we were able to meet with during the tour and to the metabolic teams at Princess Margaret Hospital, Royal Perth Hospital, Royal Children's Hospital Melbourne, Royal Adelaide Hospital, Women's and Children's Hospital Adelaide, Westmead and Westmead Children's Hospital Sydney, the Mater Hospital Brisbane and the Lady Cilento Children's Hospital Brisbane for being so generous with their time and assistance.

*Louise Healy,
Wellbeing Program Director, MDDA*

PKU Research at the Children's Hospital Westmead

We continue to make steady gains on the PKU research front.

Changing of the Guard

Sadly, Gladys Ho is leaving us to pursue higher duties – motherhood! Gladys successfully completed her PhD studies in 2013, during which she undertook comprehensive genetic studies of individuals at our NSW clinic with PKU. Her studies yielded important insights allowing us to be able to make some predictions as to the effect of specific PKU gene mistakes (mutations) on the functional capacity of the PKU enzyme. She also began work looking at the prospect of nonsense read-through therapy for PKU. We will miss her enthusiasm and great talent as a laboratory researcher, and wish her and her family all the best.

I'm pleased to report that Minal Menezes has joined the research team, hot after having finished her PhD studies on another metabolic disorder. She has a wealth of practical experience, and will be applying her talents to the nonsense read-through project.

Nonsense Read-through therapy for PAH

We have previously reported on the rationale for this approach to personalized treatment of PKU, specifically for individuals who have a PKU gene mutation that is a so-called nonsense (or stop codon) mutation. To progress this work we need to study liver cells in a petri dish culture system. As there are no commercially available liver cells with PAH nonsense mutations, we have to build these from scratch. Minal has begun genetically engineering human liver cells to create a range of nonsense mutations seen in patients. With these in hand we will then be able to test a range of potential therapeutic agents.

Progress with our GM PKU Probiotic

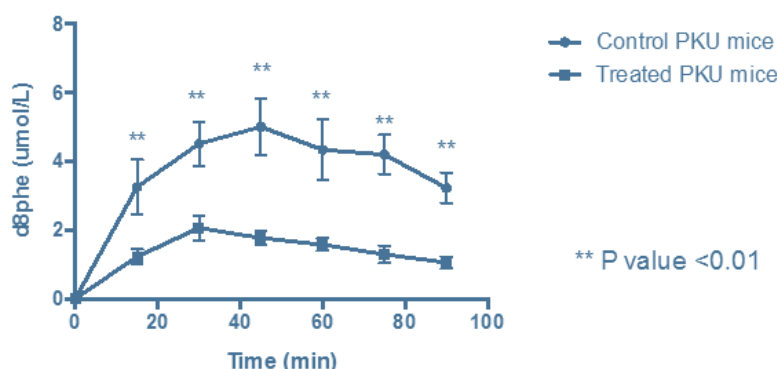
Last time I reported that we did not see evidence that our PKU probiotic (engineered to make the alternate enzyme PAL) was able to not able to reduce the amount of phenylalanine getting into the bloodstream when the PKU mice were fed protein at the same time as the probiotic. To make the GM probiotic work we have to "stimulate" it with a particular chemical in the culture broth before we feed the probiotic to mice. Following some detective work by Dr Tong, we determined that the purity of this chemical varied from batch to batch bought from the supplier, and that we needed to determine the correct dose of the chemical every time we bought a batch. Armed with that knowledge we were able to use the optimum dose to induce the probiotic to make five times more PAL enzyme. When we used the "souped up" version of the GM probiotic we now found that it was working effectively on protein, as shown in the figure. This led to a ~2/3 decrease in the phenylalanine load absorbed into the

bloodstream of the mice. This of course was very exciting, but there is still much to do.

So, where to from here? Firstly, we need to repeat this experiment, to confirm that the results are reproducible. We will then go on and examine whether we can see a sustained effect when the PKU mice are fed the probiotic over a period of several weeks. If all of that looks good, the next major challenge will be to have the GM probiotic approved by regulatory agencies so that we can move to clinical trials. This is quite a complex and tricky process, and for this we will definitely need the expertise of an organization with experience in this area to help us weave through the regulatory processes. In this regard, we are continuing our active discussions with a number of potential commercial partners.

*Prof John Christodoulou AM,
Head, PKU Clinic*

Blood d8phe in treated vs untreated PKU mice



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FSANZ Nutritional Panel Submission — Labelling changes

The MDDA became aware at the end of February that Food Standards Australia New Zealand (FSANZ) had plans to implement a recommendation to remove the 'per serving' declaration in the nutritional information panel on commercial manufactured foods. The only requirement would be for the amount per 100g or 100ml to remain.

The MDDA sent a submission as to why we did not support this recommendation.

This was submitted on the 2 March. Thankyou to all those members who completed and sent on the template form supporting our view.

The Association felt strongly that this information must remain on packaging to assist counting of protein/fats/carbohydrates. Check our website for any updates on this process that we receive.

Product Updates



Smooze Fruit Ice

Coconut & Pineapple – 0.5g protein per 100ml
 Coconut & Pink Guava - 0.5g protein per 100ml
 Coconut & Mango - 0.6g protein per 100ml
 Available from: Woolworths



Heinz Little Kids Fruit & Veg Shredz Berries, Apple & Veg

Serving size 18g
 Contains 2.0g protein per 100g
 0.4g per serve
 Available from: Woolworths



Daiya Dairy Free Cheese Shreds Cheddar & Mozzarella

Serving size 28g (1/4cp)
 Contains 3.5g protein per 100g, 1.0g per serve
 Available from: IGA stores



Daiya Dairy Free Cream Cheese Spread Plain and Chive & Onion

Serving size 30g (2Tbs)
 Contains 3.3g protein per 100g
 1.0g per serve
 Available from: IGA Stores



Jellycious Jelly — Apple & Orange

Serving size 170g
 Contains 0g protein per 100g
 Available from: Woolworths



Veg Chip — Sweet potato

Serving size 23g
 Contains 1.5g protein per 100g
 0.3g per serve
 Available from: IGA, Woolworth & Coles



The Pastry Pantry

Savoury Shortcrust Pastry Puff Pastry

Serving size 50g
 Contains 0.7g protein per 100g, 0.3g per serve
 Contains 0.9g protein per 100g, 0.5g per serve
 Available from: IGA & health food stores



Australia's Own Organic Macadamia Milk

Serving size 250ml
 Contains 0.2g protein per 100g,
 0.6g per serve
 Available from: IGA, Woolworths & Coles

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