ISSN 1440-4249

MDDA News

Newsletter of Metabolic Dietary Disorders Association

Issue 63
2017

MDDA Parliamentary Visit - Fair for Rare

On the 16th of February MDDA Executive Committee members and representatives attended the Parliamentary launch of the ***'Fair for Rare"*** campaign to highlight the importance of supporting Australians living with rare diseases ahead of ***Rare Disease Day on the 28th February.***

Mr Trent Zimmerman MP and Mr Steve Georganas MP, were pleased to co-host this barbecue, with speakers including Rare Voices Australia Executive Officer, Nicole Millis, Vice-President of the MDDA, Louise Healy, as well as Australians living with rare diseases.

Nicole Millis spoke to parliamentary members, senators, representatives from the rare disease community and guests, urging for commitment to a rare disease policy and progressing a National Plan for Rare Diseases.

MDDA Vice President ***Louise Healy*** spoke on newborn screening the impact on patient lives and the importance of early diagnosis to patient outcomes. Faye our member representative delivered a powerful patient perspective "Living with a child with a rare disease" and the impact that effective policy can have on a patient's life.

This was an invaluable opportunity to express and emphasise to Mr Greg Hunt, Minister of Health, the assistant minister Dr David Gillespise and other ministers and senators the importance of having a national co-ordinated approach to equitable access, to coordinated care, services and treatment for all Australians who suffer from a rare disease.

What can you do? Sign the 'Fair for Rare' petition and show the policy makers that all Australians living with rare disease deserve a 'fair go'.
www.fairforrare.com.au/mail-list

this issue

Donations over $2.00 are Tax Deductible

Parliamentary Visit - Fair for Rare

Following is a transcript from Louise Healy, Vice President of the MDDA, delivered at the Parliamentary Launch of the "Fair for Rare" Campaign, Parliament House Canberra, 16th February, 2017.

New born screening transforms lives….
I know this because it has transformed the life of my daughter and our family. At 4 days old Evie got a positive result from the "heel prick" test and was diagnosed with phenylketonuria or PKU. Treatment began immediately.

Had Evie been born slightly more than 50 years ago, or in another country, she would have been severely brain damaged by age 1. Today, instead of needing full time care she is a very healthy and independent 9 year old who loves reading, playing violin and practicing Taekwondo – her current ambition is to work for NASA on the international space station or to be a pop star. I am convinced that she has the capability to be whatever she chooses and that is certainly due to the opportunity that early diagnosis and intervention afforded her.

For around 50 years in Australia, most babies have been screened for a range of rare conditions including PKU, cystic fibrosis and hyperthyroidism via newborn screening providing the opportunity for early diagnosis and intervention
Interventions that can prevent catastrophic health problems including severe intellectual impairment, neurological problems, organ failure, even death – health problems that are both devastating for families and extremely costly to manage. Newborn screening reduces health care costs by preventing severe disease complications, eliminating the need for costly diagnostic tests, reducing hospitalisations and preventing intensive social welfare cost such as carer support and disability support. Newborn screening is preventative medicine at its best and is one of the most successful public health initiatives in our lifetimes.

In terms of newborn screening in Australia there is much to be grateful for and proud of. Those of us here today who are the beneficiaries of NBS feel this gratitude very deeply. *Although a positive diagnosis is confronting and presents challenges it also provides clarity, understanding, and a treatment pathway.* Without diagnosis families struggle to access care & support. Improving diagnosis must be central to making a fairer system for those with Rare Conditions.

Newborn screening urgently needs policy leadership across government. Currently, NBS is administered by the states and a lack of a consistent and transparent policy means that no new tests have been added to newborn screening since the early 2000s. Given the significant advances in diagnostic technology since then this is both shocking and totally unacceptable. It also comes at a very high cost for families whose children are born with conditions that could be screened for, as you will hear when our next speaker, Faye, shares her newborn screening story. Reliable and safe tests that will diagnose conditions that have effective treatments are available but there is currently no mechanism by which they can be evaluated and added to newborn screening panels. *For me it is heartbreaking to think that babies are dying or incurring lifelong health problems when there is a test and a treatment that can help them. As a society, as parents, grandparents, aunts and uncles we have a responsibility to provide the opportunity for every baby born in Australia to meet their full potential.*

In 2014 the need for the governments across Australia to address in this most important aspect of rare disease policy was recognised when the Standing Committee on Screening initiated the NBS working group to develop a national Newborn Screening framework that would provide:

- *clear policy guidelines for adding or removing tests from NBS regimes,*
- *decision making tools that will be able to respond to medical advances in a timely, safe and cost effective way,*
- *transparency and consistency in newborn screening across Australia.*

At the end of 2015 the proposed framework was completed. The groundwork has been done. In 2017 we ask that the framework be endorsed when it comes before AHMAC (Australian Health Ministers Advisory Council) this year and for implementation to begin. There are tests currently available that will save the lives of babies and provide many babies with the opportunity that newborn screening has given Evie and many others like her.

South Australia Gatherings

On the 16th March, MDDA held a members gathering in Adelaide where a number of families and adults got together to hear from MDDA on latest initiatives and also talk about upcoming plans and events for SA members.

Most excitingly on the cards is a national retreat to be held in Adelaide Mid 2018 - so watch this space! MDDA is setting up a SA members facebook group to keep the conversation going in this area, so if you are from SA and would like to get more involved or just keep abreast of SA happenings, please be sure to let us know.

MDDA also had the opportunity whilst in Adelaide to meet with a number of metabolic dietitians at the Vitaflo National IEM dietitians conference at Mt Lofty in Adelaide Hills. Here MDDA Vice President Louise Healy presented a workshop on Mindfulness and President Monique Cooper presented on MDDA's National Adult Wellness Coaching Program. It was a great opportunity to share and collaborate with the dietitian community and we are confident that we are building an excellent adult program that will continue to be piloted this year and eventually be rolled out to all IEM adults across Australia in 2018.

On 28 February 2017, people living with or affected by a rare disease, patient organisations, politicians, carers, medical professionals, researchers and industry came together in solidarity to raise awareness of rare diseases.
The Rare Disease Day 2017 theme '**Research**' recognises that rare disease research is crucial to providing patients with the answers and solutions they need, whether it is a cure, treatment or improved care.
The Rare Disease Day 2017 slogan is *With research, possibilities are limitless.'*
Research brings much needed hope to people living with a rare disease, their families and carers, and can be life-changing.

Due to necessity, the dynamic between patient and research is a bit different in rare disease than in more common diseases. Rare Disease Day 2017 is also an opportunity to *recognise the crucial role that patients play in research, advocacy, funding research, partnering in research projects and participation in clinical trials.*

The Genetic Support Network Victoria (GSNV) annual Rare Disease day celebration was held at the Royal Children's Hospital, Melbourne. GSNV teamed up with the Australian Genomics Health Alliance and Rare Voices Australia to showcase current research that is happening in Victoria and around the country.

Speakers included, Professor Ingrid Winship whose topic was, Rare diseases, a call to action!, Mr Frank McGuire who spoke about the Victorian Government's commitment to genetic health, Dr Katherine Howell spoke from a researcher's perspective and Gemma Brett looked at things from a clinician perspective.

The presentations concluded with a powerful emotional personal experience of living with a rare disease from Rebecca Conci . Two of Rebecca's three girls have a life threatening disease that has remained undiagnosed and has seen them spend a lot of time in The Royal Children's Hospital. Rebecca a photographer has found photographing her family's journey extremely therapeutic taking the time to capture moments, regardless of the circumstance. You can read more from Rebecca on her website www.rconciphotography.com

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Cook@Home Low Protein Master Classes

This year we have held two cook@home days. Our first one was held in New South Wales, on the 18th February and second one on the 26th March, in Victoria. Thank you to Nicholla and Simone for opening their homes to MDDA members.

There was some tasty food cooked and all those who attended picked up some handy cooking tips and certainly enjoyed themselves. Recipes will be posted on the MDDA website.

Some of the recipes included, low protein rissoles, pies and doughnuts, jam tarts, chocolate and raspberry muffins and wraps.

The cook@home days have inspired members with some great ideas, and many have made new friends as well as taking home some yummy low protein meals to enjoy.

"Thank you so much to the MDDA, Nutricia and everyone that came today!!! I had an absolute blast and would highly recommend attending or hosting your own cooking demonstrations. Thank you so much (to those that came) for the amazing day and help with all the cooking" ***Simone***

Thank you to our sponsor

NUTRICIA
Advanced Medical Nutrition

Maternal PKU

Name: Nicholla
IEM: PKU
Family: Husband – Joe & son - Max aged 1

What is the first step you must take when considering getting pregnant?
Contact your clinic and tell them that it is a step you want to take. They will help you to start the preconception diet and tell you how to do so.

How did your diet have to change when you were thinking about starting a family?
I had to go from 10g a day down to 6g a day. This was a challenge but thinking about what would happen I kept going. I had to have levels under 250 for 3 months in order to get the ok to go ahead.

What would you like other PKU women to know about the preconception diet?
It is hard, you need to be dedicated to doing it. Support from your spouse or partner is key!
Friends and family who support you is also a great help.
The days you don't feel like doing it are hardest but with a good support team it can be done.

Were there any difficulties you encountered while being pregnant and after having your baby, in regards to you having PKU?
I had terrible morning sickness. It required me to go into hospital. I struggled trying to eat food and keep it down.
My medicine was the absolute worst! I could not stand drinking it. I felt sick and would often have to start again due to sickness.
Once I passed the worst morning sickness my protein allowance started going up due to Max growing. This was great. I went up to 7g and then all the way up to 38g by the end of my pregnancy.
Once my protein got to about 15g I found it very hard. I had to eat exactly that amount of protein everyday and often did not have room to fit it in.
As it got even higher I had to start eating foods I have never eaten and it was hard. I found it difficult to eat foods as I feared I would love them and not be able stop eating them.
When it got up in the 30s I struggled more than being low protein.
It is a big change. Each week I had a blood test and my protein would go up at least 1g as Max grew a lot!

Do you have any tips for other PKU women thinking of starting a family?
- contact your clinic and let them know your plans.
- stock up on low protein foods.
- make sure you've got a support system, spouse, partner, family and friends.
- talk to other women who've been through it. They're usually happy to help.
- keep your eye on the goal and know it will eventually lead to a beautiful baby.

Tell us about your beautiful boy Max.
Max is the most incredible kid. I love him so much. He loves food, especially protein foods haha! Max tries to offer me food all the time especially when I don't have the same food as him on my plate. It's incredibly generous but he sometimes gets upset when I say I can't.
He loves both Joe and I so much and makes friends easily. We spend our days playing and learning different things. He's always a happy kid and loves to explore.

2017 Family Retreat

Friday October 6th - Sunday 8th October
CountryPlace, Kalorama Victoria

Registration is NOW OPEN!
COSTS
MDDA Members
Adults: $225.00pp twin share Adults: $285.00 single (subject to availability)
Teens 13-17: $95.00 Kids 3-12: $75.00 (incl. Sat kids club)
Kids 2yrs or under: Free

Costs are ALL INCLUSIVE and include 2 nights accommodation, all meals and activities.

MDDA retreats are designed to provide;

* relaxing weekends away
* new ways of coping
* opportunity to make new friends
* new food ideas
* a sense of wellbeing and empowerment
* educational resources
* all with direct access to selected IEM healthcare professionals/specialist presenters

Download your form NOW!
www.mdda.org.au/event/2017-family-retreat-register-your-interest-now/

Caroline's Story

The Illustrator of the book "Robin and I Explain PKU" talks about growing up with PKU, the book & changing times

My name is Caroline Thorpe I am 44 years old and I have PKU. I am on diet for life and am amongst one of the first few babies born in South Australia with PKU and put on a low protein diet from birth.

Growing up:
I remember from a young age going to see my dietician and doctor and them being very kind. My favourite part of the year was the big PKU Party held in the hospital function room, with tables full of low protein food that the hospital had organised and parents would bring a plate of food to share. Afterwards we were allowed to go into the gymnasium in the basement and play with all the equipment. There were big bouncy orange balls that we sat on and held onto the handles, rehabilitation boards that were like giant padded skate boards, and jumping on the trampolines. Many of the children enjoyed hours of these activities! I didn't actually know many other kids with PKU. When I was ten I met a lady called Gail that had PKU at one of the parties. Gail was a kind lady and was one of the first PKU ladies I had heard of that had children of her own. I stayed at her house and went shopping with her and we talked about PKU, like what points (grams) we were on and what hobbies we liked.

The book "Robin and I explain PKU": Becoming the Illustrator:
When I first went to school I started at Campbelltown Primary and I enjoyed it there. The librarian was called Mrs Brummit, (who is the author of the book) and her husband was my doctor for a while at The Adelaide Children's Hospital in the 1970's. Dr and Mrs Brummit have children and their son has PKU. I did not know that at the time. Mrs Brummit wrote a lovely children's book in the early 1980's in the year of People with a Disability *"Robin and I explain PKU"*. She sent me a copy to look at, one of the first edits, to enjoy and see if I liked and understood it. I loved it!

One day I just started to draw in the gaps above the writing. I was told there might be photos or something there eventually but Mrs Brummit wasn't sure yet. Anyway

that copy got sent back to Mrs Brummit with my drawings in it and she was told that I really enjoyed the book. Well that's when it all took off and Mrs Brummit decided to add my pictures in and she then asked me to colour and elaborate on some I had already done. I adored doing it. I had always loved to draw so that was a natural fun thing to continue with!

The book is a perfect way to explain to people young and old that don't know much about PKU what it is. At eight years old when I did the drawings I didn't know many other people with PKU. I think it was good for my family to see too, my older sister, who doesn't have PKU, read it and learnt much more about PKU. Plus it was nice for my Mum to read because then she knew other parents were going through the same thing with a child with PKU. Mum knew there were others with PKU but the book being made seemed to warrant how many people it affected! We were not the only family handling this and I think the book was reassurance of that.

Best response to the book:
Initially the best response to the book and the drawings I did were from Mrs Jane Brummit herself. The next reactions that really warmed my heart were from dieticians that told me kids were reading and enjoying the book. Even today it is still being read and being available on line it has moved with the times without changing the original input and copies! I love that kids and adults still find it relevant and fun to look at.

Helping school aged children:
I think the book helps school aged children because it is easy to read, the rhyming is cute and appealing and the drawings are relatable.
Kids draw their experiences and those pictures still reflect a lot of kids experiences today. eg playing football, taking lunches to school, having a family, having friends and for the kids with PKU the blood testing! They know looking at this they are not alone! They are like everyone else with which is what the book is all about!

Changing Times:
I hope kids with PKU that go to school enjoy all their variety of lovely low protein foods now. Many of it was not available when I was going to school or it was very hard to get. Now there are pastas, and special flours and supplements they can take to school and the cool thing is with all the different diets eg diabetics, celiac, nut & other allergies and gluten free that the PKU diet and foods just blend in with all the other diets that are around! I think we are all lucky to have such variety now.

My favourite food growing up was PKU Casserole. Mum would make a whole batch of it and there was so much left I took it to school to eat in a little container the next day. My friends had boring square sandwiches with the same filling and I had my toasted low protein bread salad sandwich and my container of casserole.

As an adult I love a good stir fry with low protein spaghetti as noodles and I love making my own spaghetti sauce that translates into a sauce for tacos filled with vegies or even nachos with vege chips!

The introduction of cooking demonstrations are such a positive helping hand to kids and adults alike, with PKU to attend them. They are fun and were not around when I was a child.

Support:
Organisations like the MDDA were non-existent for families going through things to do with inborn errors of metabolism when I was a child.

The organisation really does help and give a crucial voice to people and families with such rare conditions. It has not escaped my attention how inclusive and welcoming the MDDA is and the people whom are the back bone of it all. Thank you for not sticking your heads in the sand on such important conditions that can be forgotten about by the Government and parts of Society that it doesn't touch. You give us a familiarity, a place to mingle, you are a big help with information and an organisation that understands. Thank you!

People can still request a printed copy of the book through their dietician, and it can be viewed anytime through the link on the MDDA website under the educational resources tab.

To all those people with PKU and their families thank you for enjoying "Robin and I Explain PKU" and if you haven't yet i hope you do! *Stay healthy and be happy to all out there.*

Caroline Thorpe

Starting Primary School - with Tyrosinaemia Type 1

Name: Lachlan
IEM: Tyrosinaemia Type 1
Grade: Prep
Age: 4

Describe Lachlan's first day of school:
It went great! He was ridiculously excited about going to big boy school! (So was I lol) he ate all his lunch too which was a bonus!

What did the school/teacher know about Tyrosinaemia Type 1?
Until I told them about the condition.. nothing! They hadn't heard of it before which is understandable.

How do you explain what Tyrosinaemia Type 1 is?
I tell them that Lachlan can't break down certain amino acids in his body so he needs a special diet, formula and medication to stay alive and healthy. I also provided her with a printout of what tyrosinaemia type 1 is.

How did the school/teacher react to any special requirements?
The teacher was amazing! She was very interested to know more (which as we all know is so great!) plans were put in place so he doesn't share food and if they are having a special event in school time they call or see me to check whether it's something he can have.

Are there any major difficulties you have experienced with Lachlan starting school?
I haven't come across anything yet!

What is generally in Lachlan's lunch box
Normally grapes, banana, crackers, homemade pikelets, biocheese.

What is Lachlan's favourite food?
Grapes, crackers.

How does Lachlan manage medication/formula at school?
Lachlan usually has his formula at home so at this time I haven't had to deal with this issue.

How will you deal with class parties?
When I know there is something coming up I'll ask the teacher for a list of food that will be provided and go from there.

What is Lachlan's favourite part of school/Why?
Playground and whiteboard. He loves technology.

Any tips for future prep parents in making the transition to school easy?
Be open with them about your child's condition! Make your expectations known to them in the beginning so hopefully you don't come across any major hurdles down the road. It's a learning journey for everyone!

New Arrival
A beautiful baby boy!

Congratulations to Sasha and Steve
on the arrival of
Max
Born 22 February 2017
a brother for a very excited Elaynah!

A journey of hope: Living with Citrullinaemia Type 1

We Dodds's are a joyful family who love the great outdoors and place great value on the family unit. Our Christian beliefs have been very instrumental in travelling the family journey thus far.

We are both very hands-on parents and each day starts with reading a book with Isaac; either together or just one of us. *We try to eat as many meals together as possible, and this starts with breakfast, which is a very positive experience as it is when Isaac's appetite is at its strongest.* We both work part-time, meaning that Isaac is always with either one of us and we get to see and be part of Isaac's development. Our philosophy is that family comes first, and everything else will take care of itself as God wills it. Our almost 3-year-old son Isaac was diagnosed with Citrullinaemia Type 1 on his fourth day of life.

Diagnosis:
On the second night of life, Isaac cried for most of the night. On the following day he was lethargic and didn't open his eyes very much. When visitors saw him for the first time they commented on how placid he was (we thought this was great at the time). On day 3, we were finding it hard to feed him and he had trouble keeping warm. He was moved to the hospital's special care nursery to warm up under the lamps, but his condition worsened fairly rapidly with blotches on his lungs showing on x-rays, heart problems and trouble breathing. He was grey in colour. The NETS team came, and once stabilised, he was transferred to Sydney Children's Hospital ICU. Sarah needed her Obstetrician to discharge her so she could travel with Isaac and overhearing the call in the background was his wife, who is a geneticist at Sydney Kids. What a God send. She was the on-call doctor that weekend so she advised straight away what tests should be done as she suspected a metabolic issue and immediately to meet us in ICU.

With major organs affected, the medical team gave us "that talk" with a few possibilities of what he might have and "if" he survived he would most likely have significant brain damage. They were concerned that we didn't understand what they were saying but we just responded with "God is in this". Early the next morning the medical geneticist at the hospital came back to us with a diagnosis. During this time, Isaac's ammonia reached 860 and he required dialysis to filter his blood of the ammonia. He also developed a swallowing problem with aspirations so had an NG tube for all feeds. The NG tube was no longer required after 10 months.

Management:
The management of this condition involves a combination of low protein and high calorie diet and medication, both of which will be in place for the rest of his life. The diet allows Isaac to consume approximately 1g protein/kg of body weight/day. The medication, L-Arginine Hydrochloride and Sodium Benzoate, combine to soak up harmful ammonia levels in his urea cycle, as well as driving the cycle to be more efficient and get rid of the ammonia. He has these 3 times per day.

We are very good at systemising meal times and also prepare all medication for the day at the beginning of the day. He is a very fussy eater as a toddler. Isaac really enjoys Weet Bix, and epitomises the "Aussie kids are Weet Bix kids motto." He also loves porridge and nuts, and the odd chocolate treat.
Because we are systemised and both very hands-on, it has become part-and-parcel of any outing to prepare an assortment of Isaac's foods. In this way he is able to choose from a selection of foods; something he likes to do as it gives him control over what he eats. Life is also made simpler through the use of the Easy Diet Diary app. This is an app that helps to calculate protein levels in everything Isaac consumes. It lists protein for each meal and gives a daily total. Food can be entered either by scanning a bar code, typing in well-known food types or brands, by

adding in own food, or by using the 'recent' function. And each of us can log in from our devices.

We really enjoy the outdoors, so a trip to the local park, where Isaac can ride his scooter and run around, is the way to go.
It is always a challenge when Isaac has a runny nose or a temperature. We are not able to present to our local hospital; rather we travel to Sydney Children's Hospital to have an ammonia check done. And this is not as simple as a finger-prick test; this involves a full blood-count. That is the nearest place that tests ammonia and also stocks the IV medications he would need if the result is high. There are certain symptoms we must look out for; many of which are exactly the same as common harmless bugs (for other kids) like lethargy, high temperature, vomiting, diarrhoea. We are always mindful of Isaac playing near children who have sniffles as this could cause him to become unwell, and are very vigilant about not allowing Isaac to share any foods that other children may offer him.

Educating others:
We find that educating others about Isaac's condition is most effective in a conversational situation, where people naturally seek information on how he is doing. Every time we share this information it helps us, as parents, to better understand the reality of the condition. On one occasion a lovely older lady at a church group almost gave Isaac a chocolate biscuit, so in groups we often remind everyone on a regular basis that any food has to be checked by the parents before being offered to Isaac.

We would like other families to realise that living with this condition can be a totally manageable affair. At first it seemed doom-and-gloom, but we had the attitude that we were going to do whatever we had to do so that our son would lead a relatively 'normal' life, whatever that meant. *We are now living the wonderfully rewarding experience that is watching Isaac grow and develop.* He has no signs of any developmental concerns to the Neurologist's amazement. He is a joy-filled, determined, intelligent little boy who seems to know he has a point to prove. He is testament to the power of belief and living against the odds.

As we have said above, living with an IEM can become "living to the full". It is all in the attitude we have and the hope we see in Isaac. *Just keep doing what you need to do and enjoy the journey.* It is such a blessing to be able to take that journey together.

Sarah & Grant Dodds

Fundraising Updates 2017

Protein Challenge Fundraiser

We need YOU to help kick off a major awareness & fundraiser campaign to commence on 1st May, marking the beginning of PKU Awareness month 2017.

Announcing the launch of **"How Low Can You Go"** the **Great Protein Challenge** – we want you to challenge your family, friends, work colleagues, school communities and sporting clubs and your local Federal MPs. The objective of this challenge (similar to the 40 hour famine) is to provide people with a way to experience what it might be like to have PKU (by following a strict PKU style low protein diet for a day) and being educated on the complexities and many challenges that it imposes on individuals faced with managing PKU on a daily basis. Participants can choose to Pledge or Play.

Pledge your daily protein intake for one day or
Play – design your daily meal plan so your entire food intake for the day does not add up to more than 10 grams of protein for the day.

This fundraising event will raise funds which will directly enable our organisation to continue delivering our programs and services to families.

Donations and funds raised can be made at www.givenow.com.au/mddahowlow

GiveNow
Give more, give smarter, give now!

Entertainment Books

Thank you to **Tracey Scott** for raising $259.00 from the sale of the Entertainment Books 2016.
The funds raised by Tracey will be donated towards PKU research at The Children's Hospital Westmead and to the MDDA .

Tracey will be co-coordinating the sale of the Entertainment Books again this year for the same cause. So jump on board early to make full use of the offers. See our information page for website details.

Thank you to the Hellings family for raising funds and awareness for the MDDA

Yarra Glen Bowling Club

Recently we had the opportunity to play lawn bowls amongst the community of Yarra Glen to represent the MDDA and Yarra Flats Baking Co. The tournament was held over 12 weeks. Thank you to Yarra Glen Bowling Club for holding these charity bowls events and their donation of $200 to the MDDA.

I would also like to say a big thank you to my work, Yarra Flats Baking Co at Yarra Glen and Mount Dandenong, for supporting my daughters with PKU, nominating the MDDA as their charity to play for and helping raise awareness of PKU and other metabolic disorders.

Yay team!
Sonia Hellings

.

MDDA were saddened to hear Isaac Gravina passed away January 23 at age 21 months. Isaac and his twin sister Charlize were diagnosed with the rare metabolic disorder Propionic Acidemia (PA). Propionic Acidemia affects one in 250,000 births in NSW and is an inborn error of protein metabolism (IEM).

A Go Fund Me campaign has been set up to raise funds for the Genetic Metabolic team at The Children's hospital at Westmead to fund more resources, research and treatments for diseases to help save Charlize and ultimately save more children living with an inborn error of metabolism.

You can donate to the campaign at **www.gofundme.com/baby-charlize-vs-propionic-acidemi**

You can follow Charlize & Isaacs' journey on their facebook page **Isaac & Charlize's Journey with Propionic Acidemia and Liver Transplant**

We send our love and thoughts to Julie, Paul, Amelia and Charlize

Information Updates

WHY YOUR MEMBERSHIP MATTERS

⇒ 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.

To further involve yourself with any of our initiatives listed above please contact us. We rely on the passion and generosity of all our volunteers to continue to work for the benefit of all members and the IEM community.

Entertainment Books

Purchase your entertainment book and support the MDDA at www.entbook.com.au/91531w0

The entertainment memberships are available in two different formats;

Entertainment Book, - offers are redeemed via Entertainment Gold cards and vouchers
Entertainment Digital Membership -allows you to redeem offers directly from your iPhone or Android smartphone.

Thanks to Tracey Scott for organising.

IEM BABY BEARS

Thank you to Emily Large for kindly knitting some MDDA bears for us to give to newly diagnosed infants.

Emily's grandchildren who have PKU are now in their twenty's. It is wonderful to have the support from the Large family over the past 20 years.

IEM GRANT

Just a reminder that to remain eligible for this grant you have individual responsibilities such as maintaining your diet as evidenced by regular blood tests and keeping clinic appointments. Failure to comply may mean your access to the grant could be removed.

Payment Schedule for 2017

The IEM food grant payment for 2017 is due by the Friday of the first full week of each month. Payments due to arrive by:

7 April, 12 May, 9 June, 7 July, 11 August, 8 September, 6 October, 10 November, 8 December

For questions please contact the Department directly on (02) 6289 8980 or iemprogramofficer@health.gov.au

PKU Direct Oceania is a new company providing Promin products to the Australian market. They import low protein foods from the UK.

More information can be found at:
www.pkudirect-oceania.com

PKU Direct Oceania

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Ph: 1800 367 758

PKU Direct Oceania - www.pkudirect-oceania.com

Platypus Foods – www.platypusfoods.com.au

Moving or changed your email—don't forget to notify MDDA! email office@mdda.org.au

PKUNSW UPDATES

Running for PKU

On Saturday April 8th Kate is running 12km through Ku-ring-gai National Park, New South Wales and hoping to raise some money and awareness for PKU.

Kate has two nephews with PKU. To read more about her story or to donate go to:
www:give.everydayhero.com/au/running-for-pku

Lab coat awareness walk - May

Come along and join in our Lab Coat Awareness Walk. We walk in our lab coats with our team mascot Phen and spread the word about PKU. Get a TEAM together, walk by yourself, your cat or dog in your Lab coat to promote awareness of PKU around Australia.

Membership

PKUNSW Memberships are now up for renewal go to:
www.pkunsw.org.au/get-involved/membership

For more information and confirmation of dates for any of these events go to
www.pkunsw.org.au or
email: info@pkunsw.org.au

City2Surf

How else to get more involved and also promote PKU than to run or walk the Sydney City2Surf in August with our team mascot 'Phen' the platypus. Details of how to join us or register are available on our Events page from June.

Gala Ball

Usually around June each year we hold a a Gala ball. This our major fundraiser to raise funds into research currently ongoing into PKU. Tickets always sell fast to this extravagant event so you had better book them early.

Early Career Researcher Kick Start Grant Update – Dr Minal Menezes

Nonsense suppression as a treatment for phenylketonuria

This study aimed at investigating nonsense suppression as a potential treatment for Phenylketonuria (PKU) caused by mutations in the phenylalanine hydroxylase (PAH) enzyme. PKU is recessive disorder affecting 1 in 10,000 newborns in Australia).

One of our primary aims was to determine the most effective nonsense suppression agent (drug which restore a functional enzyme) for phenylalanine hydroxylase (PAH) nonsense mutations in an *in vitro* model. Based on our assays and toxicity information available, a drug named Amlexanox, which has been clinically approved for many years and has been used for its function as an anti-inflammatory antiallergic immunomodulator was chosen as the nonsense suppression agent of choice.

Since the proximate cause of PKU is deficiency of PAH in liver, we aimed to test the efficacy of nonsense suppression in human liver cells (Hep3B). However initial studies revealed that Hep3B did not express PAH enzyme levels which could be detected by mass spectrophotometry. Treatment of the liver cells with the co-factor tetrahydrobiopterin showed increase in PAH activity; however when the concentration was increased it was a bit toxic to the cells. Previous studies have shown that addition of sepiapterin produced a 4-fold enhancement in PAH activity in hepatoma cells and we are in the process of replicating those results.

One of our key objectives was to investigate whether the nonsense suppression drug selected as the most effective nonsense suppression agent in our invitro studies has the capacity to restore the PKU phenotype in PKU "nonsense" mice. We have now collaborated with A/Prof Cooper (Deputy Head of the INMR) whose laboratory has expertise in using CRISPR/Cas9 technology to generate mouse models, which will now assist us in this crucial step of the study.

The work from this study has been presented at the Journal club at Kids Research Institute and PKU Rotary Association NSW.

Autumn Recipes

Tomato Onion and Olive Risotto

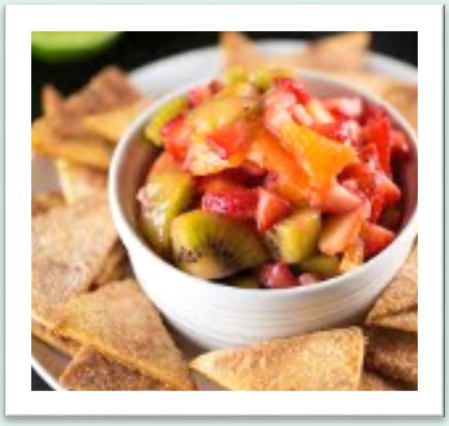
(Calculate all recipes as per guided by your dietitian)

Ingredients
1 tablespoon butter
1 tablespoon olive oil
1 onion chopped
1 clove garlic crushed
75g low protein rice
½ cup tinned chopped or crushed tomato
6 Kalamata olives sliced
6 sun dried tomatoes chopped
1 cup chicken stock (1 tsp chicken stock powder + 1 cup water)
Fresh basil leaves for garnish if desired

Method
1. In a non stick saucepan heat butter and oil until butter is melted, add onion and garlic and cook until soft.
2. Add rice and cook for 2 to 3 minutes.
3. Add all other ingredients and stir to combine.
4. Reduce heat to low, place lid on saucepan and simmer approximately 20 minutes, stirring 2 to 3 times during cooking.
5. Garnish with basil and serve with salad or roasted vegetables

If Risotto is still too moist, take lid off and simmer, stirring until thick.

Fruit Salsa and Cinnamon Chips



Ingredients
2 kiwi, peeled and diced
2 apples, cored and diced
1/4 cup raspberries
2 cups of strawberries, diced
2 tablespoons sugar
1 tablespoon brown sugar
3 tablespoons jam of choice
10 low protein pita pockets
Cooking spray
2 tablespoons cinnamon sugar

Method
1. In a large bowl, gently toss together the apples, raspberries, strawberries, sugar, brown sugar, and jam.
2. Cover and chill 15 minutes minimum.
3. Preheat oven to 180 °C. Spray each side of the pita pocket and cut into wedges.
4. Arrange in a single layer on a large tray lined with baking paper. Sprinkle chips with cinnamon sugar.
5. Bake in oven 8 -10 minutes or till crisp.
6. Repeat with remaining pita pockets.
7. Allow to cool 15 minutes and serve with chilled fruit dip.

Chocolate & Raspberry Muffins

Ingredients (Makes 12)
2 1/2 cups low protein flour
3 teaspoons baking powder
1 cup brown sugar, firmly packed
2 teaspoons cocoa
90g butter, melted
1/2 cup oil
2 teaspoons vanilla essence
200ml low protein milk
1 cup raspberries (fresh or frozen)

Method
1. Preheat the oven to 180 °C. Mix the low protein flour, baking powder, brown sugar and cocoa together.
2. Mix the butter and the oil together, add the vanilla, low protein milk, and add to the dry ingredients, mixing with a wooden spoon.
3. Add the raspberries and mix well without overbeating.
4. Spoon the mixture into a muffin tin lined with patty pans and bake for 15 minutes or until cooked.

Product Updates

Vege Deli Crisps - Chia
Protein per 100g: 2.7g
Protein per serve: 0.5
Serving size: 20g
Available: Coles, Woolworths

Organic Matters - Vegan Bacon Bits Original
Protein per serve: 0.5g
Serving size: 7g
Available: Coles, Woolworths

Um - Vegi Crisps Carrot
Protein per 100g: 3.1g
Protein per serve: 0.37g
Serving size: 12g
Available: Woolworths

Oriental Fields - Crunchy Rice Rolls
Protein per 100g: 2.5g
Protein per serve: 0.3g
Serving size: 11g
Available: Costco

Eskal - Vanilla Tea Biscuits
Protein per 100g: 1.1g
Protein per serve: 0.3g
Serving size: 28g
(4 biscuits)
Available: Coles, Woolworths

Orgran - Outback Animals Chocolate Cookies
Protein per 100g: 5g
Protein per serve: 0.8g
Serving size: 17g
Available: Coles

Mrs Macs - Gluten Free Curry Vegetable Pie
Protein per 100g: 1.1g
Protein per serve: 2.0g
Serving size: 175g
Available: Coles, Woolworths

Eskal - Original Cones
Protein per 100g: 0g
Protein per serve: 0g
Serving size: 4g
Available: Coles, Woolworths

Nudie Coconut Yoghurt - blueberry
Protein per 100g: 1.3g
Protein per serve: 1.1g
Serving size: 85g
Available: Coles, Woolworths

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MDDA News

This newsletter will only grow and get better with your input. Please share your stories, ideas and tips, birthday celebrations and functions with us.

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