



# MDDA NEWS

Newsletter of Metabolic Dietary Disorders Association

Issue 55  
2014



## Raising IEM Awareness

In this issue we celebrate raising IEM awareness within our community with many wonderful events organised by our members to celebrate **IEM Awareness Month** in May.

We go on a tour through the **Royal Children's Hospital Newborn Screening Lab** and see first hand just what happens to those Guthrie Cards at the other end of the mail run. We had a great turnout of families and little ones sharing in the adventure during Victoria's 0-5 year old get-together in July (the full story of the event can be found on page 6).

In October we have our bi-annual **Family Retreat** taking place at the **Sydney Conference & Training Centre, Ingleside, NSW**. This will be our 6th family retreat held and is shaping up to be our largest yet. So if you haven't registered be sure to do so and join in the fun of meeting fellow members, sharing stories and learning from healthcare

professionals speaking at the retreat. PKU advocate **Kevin Alexander** will be joining us from the United States to share his story and film the experiences of IEM individuals here in Australia.

MDDA member **David Contera** gives us a wonderfully detailed account of his time at the **ESPKU Conference** in Antwerp as well as an informative tour through Dr Schars in Spain, a medical nutrition group manufacturing low protein products.

You will find some fantastic low protein recipes to ward off the winter chill as we patiently await the return of the warmer summer months. There are also some great new products included on the back page which have been shared by our members through our MDDA Facebook site.

*Pictured above - families attending the 0-5 year old get-together at the Royal Children's Hospital in July*

## this issue

PKU Awareness Month	P.2
Eden Kate Drewitt	P.3
Far North Queensland Clinic	P.4
Newborn Screening Lab Tour	P.6
What's that lab?	P.7
Winter Recipes	P.8
ESPKU Conference 2013 - A Fathers Perspective	P.10
Update from The Children's Hospital Westmead	P.13
Introducing Kevin Alexander	P.14
Tell us your IEM stories	P.14
NSW Family Retreat 2014	P.15
Product Updates	P.16

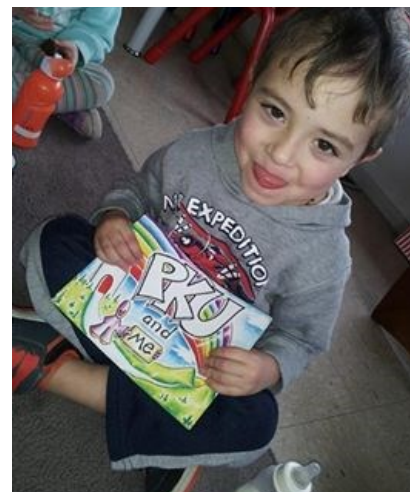
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# PKU & IEM Awareness Month - Reef Emmerson



In past issues we have travelled along with Reef Emmerson, sharing the ups and downs of his PKU journey during his first and second years. On the 1st May this year Reef Emmerson's crèche celebrated PKU awareness day by reading 'PKU & Me' for story time.

It is great to see Reef again (pictured right with a wonderful cheeky smile) sharing in his milestones, and it's lovely to see him spreading the word about PKU with his teacher and crèche friends to celebrate PKU awareness month.



## PKU Awareness Month - Playing for a PKU Cure

Regional Victorian members Justine Inglis and Nikki White put their efforts into raising awareness for PKU in May, through holding a fundraising and awareness event at the Happy Valley Hotel in Ovens on Saturday 3rd May 2014.

Justine Inglis lets us know how the event unfolded...

The idea for a PKU fundraiser came to me when trying to support a fellow PKU adult in Bendigo Victoria who was also wishing to do a fundraiser but finding it difficult to raise interest and support and put a shout out for some help on Facebook.

I thought mmm!! why not do one locally in Myrtleford where I live also! so the idea for a "Playing for a PKU cure" took shape.

I am thankful I knew a local musician and fellow nurse - Tracey Hamilton who supported me along with my partner Kev Black and his friend blues artist musician Willie Golightly who travelled from Gippsland to play for the fundraiser.

Without the artists the event would not have been such a success. I am very grateful for all their kind donations of money, time & talent! Also a big thanks to my mum

Gaewynne Old for helping me to set up the event.

The event commenced at 3pm with Local musician Terry Lockwood, then Tracey Hamilton, Simone and friends and Rudi Katterl, finishing with Willie Golightly.

The event raised \$411.00 in donations from raffles throughout the evening and donations at the door. A fab night was had by all and I also met another family who live in Porepunkah with three children two of whom have PKU! Throughout the evening the kids helped with drawing the raffles (Violet Anders 5yrs is pictured helping below) and danced all night long! I was grateful to meet up with a local family also on the PKU journey.

I am really hoping to do it all again next year!

The online fundraiser <https://give.everydayhero.com/au/juz-nikki-pku-fundraiser-2014> has raised \$1,682.55.. not too shabby for a first time effort!

A big thank you to all who supported me & the "Playing for a PKU cure" event especially the local Myrtleford businesses.

*Justine Inglis*





# Welcome Eden Kate Drewitt



Our very own Vice-President, **Richard Drewitt** and Secretary, **Katy Drewitt** have some very exciting and much anticipated news to share with our members. Their beautiful daughter **Eden Kate Drewitt** has made her arrival!

Our pictures show a very proud mum and dad in the hospital with Eden and a comparison of mum and bub heel prick cards. To the right is Eden with a lovely big smile on her face showing just how happy she is to be here and a not so happy Eden having her heel prick test.

Katy and Richard have eased into parenthood exceptionally well and couldn't be happier. Here are a few words from Katy on her new role as a mum...

"After many weeks, months and even years of planning and hard work, Richard and I welcomed our beautiful daughter Eden Kate Drewitt into the world on 2nd June 2014 at 2:59pm, weighing in at 7lb 3oz.

As I said this has been years in the making, involving losing 40kgs, getting levels under control for pre-conception diet, and then managing the PKU Maternal diet once we found out we were pregnant.

We are so thrilled about becoming parents and Eden couldn't be more perfect. "

*Katy and Richard Drewitt*



## Show Your Colours Day

**A big thank you to St Mark's Anglican Community School for your donation of \$1000.00**

The schools annual Show Your Colours Day was held on 27th June to help raise funds and awareness for those living with IEM's

Students were encouraged to wear their favourite sporting team colours. On the day the school was a sea of colour. Furthermore the Duke of Edinburgh students cooked 250 cup cakes adorned in different team colours. They were sold at recess time and were a great success.

It sounds like it was a great day for all students and the money raised will be of great benefit to help support those living with an IEM



**facebook**

**Calling all UCD, GA1/GA2, MSUD, MMA, MA, OTC & all other IEMs**

Join the discussions on our Facebook group  
[www.facebook.com/groups/metabolicdietarydisordersassociation](http://www.facebook.com/groups/metabolicdietarydisordersassociation)  
and request an add to the group



# Far North Queensland Clinic



**R**yan and Kim Large represented the MDDA once again for the Far North Queensland Clinic held in Townsville and Cairns over the 27th to 28th May 2014. Here is Kim's story...

I am a mother of 6 children. **Ryan** and **Zane (25)** both have PKU, **Kyle (23)**, **Cory nearly 21**, **Jess (17)** and **Nikaela (16)** and a wonderful husband **Roy** (can't leave him out).

As most of you know having a child with something different to others makes life a little different and full of many ups and downs... also fun and rewarding times. I've had many of these over the years, many people (and often those

most closest to you) don't really understand what a day in your shoes is all about or your child's.

My boys have made me a stronger, more open person and I'm truly blessed to have my 6 kids in my life to teach me many of life's lessons...often some I'm not sure I want to learn!

Once again it is my pleasure to go on clinic and to share with you some of our special times...

My daughter Jess and I drove to Townsville on Sunday before clinic for the night to catch up with **Dr Jim**, **Nurse Anita** and other families for dinner at Townsville. It was a good chance to relax and chat with everyone, some old friends and some new friends. Jess

enjoyed putting faces to the names that the twins and I often talk about.

We drove back home Sunday night just before midnight, long drive (3hrs) but well worth it. Ryan and I started our journey Monday night from Proserpine and headed back to Townsville for clinic on Tuesday morning...yum fruit, chips and lollies for the drive.

The Townsville crew are **Jamie-Leigh(19)** studying at Uni & **Luke Denison** year 11, **Melissa Smith (27)** PKU with a lovely baby boy **Cooper (8mths)**. She told us a little about her pregnancy and how she went. Looking at Cooper she did a wonderful job.

*(Continued on page 5)*





## Far North Queensland Clinic cont

Big brother **Angus (8)** PKU has a new baby sister **Emma** and a sister **Lucy (6)**. Angus loves pasta, spring rolls, fried rice and is a fantastic big brother. **Mia (3)** PKU goes to kinder now and loves gnocchi and enjoys her PKU coolers. It was great to see awesome **Kurtis (7)** PKU from Charters Towers, he is one of Ryan's little favourites. Young **Lucas (5)** enjoys coming to clinic with his mum.

The Acton family is growing with mum Sarah due to have baby number 3 in a few weeks (how exciting). Their son **William (16mths)** PKU is a ball of energy. **Troy** was at clinic chatting about which foods he likes to eat and what he gets up to in his spare time after work. **Stephanie Butler (25)** PKU made the drive up from Ayr.

We meet a lovely family that had just moved to Townsville from Toowoomba, **Erin Crause (14)** and her dad. Erin loves video games, dogs, music and drawing. **Arle McGregor** and **Lee Mayfield** we missed seeing this year as we had to catch our plane to Cairns for Wednesday's clinic.

The Cairns crew this year are **Viesha (10)** who has the most adorable smile. **Javan (5)** and his mum. Javan spent most of his time playing in the enclosed play area, he loves going to school and he is going to try the new protein bars. **Shari (19)** loves catching up with Dr McGill.

The Howard family is always a pleasure to catch up with as they hold a dear spot in our hearts as we first met the Howards in Mackay many years ago. **Emily Howard (8)** PKU.

We meet another new family to the Cairns area, **Tonia Savo** and her 2 children **Lily** and **Caleb** who both have PKU. There were

a couple of other families at clinic, but they came in fast and left just as quick so Ryan and I didn't get a chance to chat to them.

Another great reason to come to clinic is not only to see your Doctor and to catch up with other wonderful people with the same disorder as you, but also to sample all the new and improved products that are coming onto the market and making our lives a little easier.

Being up here in the Far North most of us only go to clinic once a year and some of us have phone conference calls, so clinic time is pretty special. It's the only time a lot of us get to see each other, swap stories, laugh, cry together and have lots of fun.

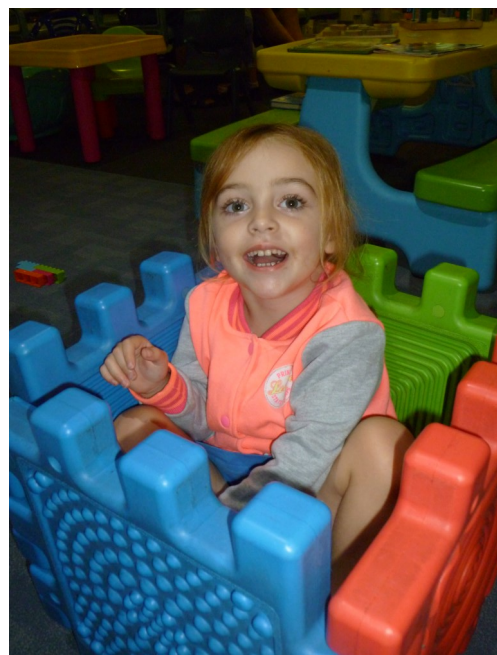
Ryan and Zane are 25 this year and every year they go to clinic they learn something about themselves and other families and how truly blessed we all are to have each other and to be able to help someone else in some small way. It's great other kids getting to see someone as special as them and knowing they and their families are not alone.

Zane works in Mackay as a fitter and turner and goes out to the mines sometimes.

Ryan works in Proserpine with his dad at a truck company as a fitter and turner. Both boys have long term girlfriends. And the boys still let me jab their finger for a blood test sometimes (which I secretly enjoy).

Well that's all from me again for another 12 months, This year is just rolling on...Santa is not that far away..

Kim Large





# 0-5yr Old Catch-up

## Visiting the Victorian Newborn Screening Lab



It is vital to teach our children nice and early about their 'special tummies' and an exciting opportunity came up for us to give our little ones a tour of the Victorian Newborn Screening Lab to see exactly what happens to those Guthrie Cards that cause those 'ouchies' each week!

On Tuesday 22 July 2014, Wendi Emmerson arranged for MDDA members to meet at the Royal Children's Hospital for lunch in the Family Resource Centre. It was great to see so many attend.

Mia and her mum & dad, Alison and Steve travelled from Moe, little Jensen, his mum Michelle and Nanna travelled from Geelong and big Jensen, Crystal and their mum Cat made the trip from Yarrawonga. We were also joined by Reef, Lakeyn, big Chloe, little Chloe (who celebrated her first birthday with us!), Lachlan, Sarah, David, Harry, Dane and their mums.

After lunch and getting to know one another we headed off to visit the Meerkat enclosure and the Aquarium the RCH are lucky to have onsite!

We were then greeted by Tony who took us up to the lab (in the midst of a fire drill). It was so interesting to see what happens to the cards once we send them off in the post. The lab performs all the dried blood spot (DBS) tests for children & adults with an IEM in Victoria & Tasmania.

Tony who has been working in the lab for over 20 years talked about the process from the DBS on the 'Guthrie Cards' through to the results

being sent to our clinic and in turn us being notified of our levels.

He showed us the machine which punches out 3 tiny discs from one blood spot to the Tandem Mass Spectrometry which analyses and reports the results of our levels to their computer.

Can you believe there is a PKU adult working in the lab? Tony said every day for the staff it's a constant reminder of how important Newborn Screening and our blood samples are.

Tony and the lab staff were so welcoming and helpful. A few of us hand delivered our cards which they gladly accepted, nothing was too much trouble. It is nice to know that these wonderful people working away in the lab actually care about the work they are doing.

Our thanks go out to all at VCGS and especially Tony. We hope they will have us back and would love to make this a regular visit.

A huge thank you to Wendi Emmerson for organising the day. We really appreciate your time and effort in arranging our get-togethers.







## What's that lab?

**W**hat's that lab?, aims to inform our members about the laboratory services of the Victorian Clinical Genetics Services (VCGS). In this edition James Pitt, Head of Newborn and Metabolic Screening Group, tells us about the lab's role.

Each working day, an average of 300 samples from Victorian babies arrive at the Newborn Screening (NBS) Laboratory at VCGS. The lab tests >99% of babies born and samples are collected in maternity hospitals, large and small, across the state. A few drops of blood are taken from the baby's heel between 2 and 3 days of life and placed on a special absorbent paper card, often called a "Guthrie card" after the inventor of this system. These dried blood spots, and the ease with which they can be transported, are the keys to effective newborn screening programs across the world. Samples undergo three NBS tests:

- Thyroid stimulating hormone, a marker for congenital hypothyroidism
- Immunoreactive trypsinogen, a marker for cystic fibrosis
- Tandem mass spectrometry testing, which detects 25 genetic disorders affecting

metabolism, phenylketonuria and medium chain acyl-coenzyme A dehydrogenase deficiency being the most frequent.

The good news is that these disorders are rare. Overall, 1:1,000 babies are affected by one of the above disorders. The other good news is that effective treatments are available for most of them. The important thing is that treatment is most effective if started early, preventing the inevitable long-term damage to the baby that would otherwise occur. Since it started in 1966, the NBS program has prevented the major disablement or death of hundreds of Victorian babies, allowing them to develop into normal, healthy adults.

***This article has been reproduced from the GSNV's Winter 2014 Newsletter on permission from the Genetic Support Network of Victoria***

For further information see the VCGS Newborn Screening website <http://goo.gl/8tx85j>  
Some recent publications:

- Charles T., Pitt J., Halliday J., Amor DJ. Implementation of written consent for newborn screening in Victoria, Australia. *Journal of Paediatrics And Child Health*. 50(5):399-404 (2014)
- Massie RJ., Curnow L., Glazner J., Armstrong DS., Francis I. Lessons learned from 20 years of newborn screening for cystic fibrosis. *The Medical Journal of Australia*. 196 (1) : 67 - 70 (2012)

# Warm Winter Recipes

## Minestrone Soup

### Ingredients

- 3 large carrots, roughly chopped
- 1 large onion, roughly chopped
- 4 celery sticks, roughly chopped
- 2 large sweet potatoes, roughly chopped
- 1 small turnip, roughly chopped
- 100g mushrooms, roughly chopped
- 100g cabbage, shredded
- 1 tbsp olive oil
- 2 garlic cloves, crushed
- 2 tbsp tomato purée
- 1.5 litre vegetable stock
- 400g can chopped tomatoes
- 150g Loprofin Long Spaghetti uncooked, snapped into short lengths
- 1 packet of Loprofin Part Baked Rolls

### Method

1. Heat the oil in a large saucepan and fry the onion, garlic and mushrooms over a high heat for 5 minutes, until softened.
2. Add in the celery, sweet potato, turnip, tomato purée, stock and tomatoes and bring to the boil.
3. Reduce the heat, cover and simmer for 10 minutes.
4. Add in the Loprofin Long Spaghetti, then cook for a further 10 minutes, adding the cabbage for the final 2 minutes.
5. Season to taste and serve with Loprofin Part Baked Rolls.



## French Style Lasagne

### Ingredients

#### Sauce

- 390g can tomatoes
- 4 tbsp vegetable stock or water

#### Lasagne

- 1 x 200ml carton Low Protein Milk
- 15g butter
- 15g Loprofin Low Protein Mix
- 1 tsp French wholegrain mustard, optional
- salt and pepper to taste
- 6 sheets Loprofin Low Protein Lasagne
- 2 x 12.5cm x 10cm ovenproof dishes or foil containers

### Method

1. Pre heat the oven to 200C
2. Combine the tomatoes and stock in a medium sized bowl
3. Pour the Low Protein Milk into a pan, add the butter and heat gently until melted. Whisk in the Loprofin Low Protein Mix.
4. Bring to the boil, stirring briskly to achieve a smooth sauce. Stir in the mustard and adjust seasoning to taste.
5. Spoon about a third of the tomato mixture over the base of the two ovenproof dishes. Cover each with a layer of Loprofin Lasagne sheets, breaking the sheets to fit the dishes.
6. Repeat the layers, finishing with lasagne sheets, then cover with the sauce.
7. Place the Lasagne in a preheated oven for 25 minutes, until the top is golden brown. If desired serve with a crisp salad

Serves 2

Tip: Keep the lasagne sheets in a single layer, so that they will cook evenly, in contact with the sauce.





# Warm Winter Recipes

## Winter Vegetable Stew with Dumplings

### Ingredients

#### For the stew

200g onions  
150g carrots  
100g parsnips or turnips  
100g sweet potatoes  
100g celery  
2 tbsp oil  
1 tbsp tomato puree  
1 Tin chopped tomatoes  
200ml water  
salt & pepper  
suitable gravy browning

#### For the Dumplings

75g Fate All-Purpose mix  
25g margarine  
35ml water  
extra Fate All-Purpose Mix  
salt & pepper



### Method

1. Chop the onions, carrots, parsnips or turnips, sweet potatoes and celery into even sized pieces. Heat the oil in a pan and gently heat. Cover the pan with a lid and leave to cook for 5-10 minutes until softened and lightly browned.
2. Stir in the tomato purée, tin of tomatoes & water.
3. Season with salt and pepper and add a few drops of gravy browning to give a rich colour.
4. Bring to the boil and then turn down the heat. Leave to simmer gently for 20-30 minutes until the vegetables are cooked.
5. Meanwhile make the dumplings: Place the Fate All Purpose Mix into a bowl and rub in the margarine until the mixture resembles fine breadcrumbs. Stir in salt and pepper and add the water
6. Using a metal spoon, mix well until the mixture is smooth. (It should be just like a soft pastry dough).
7. Take about a teaspoon of mixture and using a little extra All-Purpose Mix on your hands, roll into a ball. Put the shaped dumpling on a plate while you shape the rest of the dumpling mixture.
8. To Finish: Make sure your low protein stew is cooking and simmering well.
9. Take the dumplings and gently place them on the top of the stew in the saucepan. Do not stir in. Put a lid on the pan and leave to cook for about 5 minutes.
10. They will have risen and be light and fluffy. After this time, it is a good idea to take a dumpling out of the pan and cut it in half to test that it is cooked.

## Fate Pancakes

### Ingredients

200g Fate All-Purpose Mix  
1/2 tsp baking powder  
pinch salt  
75 ml oil  
450 ml water  
A few drops of yellow coloring

### Method

1. Place the Fate All-Purpose Mix into a jug and stir in the baking powder and salt.
2. In a separate jug, measure the oil and water and add a couple of drops of yellow colouring.
3. Using a fork or whisk, quickly stir in about half of the oil and water mixture. Stir until creamy, then add the rest and stir well until blended. The mixture will be quite thick.
4. Place a frying pan on a low heat until warm. Do not add any oil.
5. Add about 2-3 tbsp of the pancake mixture and use the back of a spoon to gently spread the mixture to the edge of the pan.
6. Leave to cook for about 1 minute or until the top is dry and the base is brown. Turn the pancake over and cook for a further minute or so. Place onto a plate and keep warm while you cook the remaining pancakes.





# ESPKU Conference 2013 - A Fathers Perspective

I took the opportunity to arrive in Antwerp a day before the conference. The extra day would allow me to get over the jet lag and allow my body to adjust to the time difference. So I thought!

I arrived in Brussels and took the train to Antwerp. I was really excited and looking forward to the exchange of knowledge and experiences that the conference would bring. Do patients and their families in Europe do things differently to us back home in Australia, how does the ESPKU community rally together, what are they doing differently? These and many more questions I would soon hope to answer.

*As a father with two PKU children, the main purpose of this trip would be to network with members of the PKU community and meet other parents in the situation Maria and I find ourselves in.*

Once I arrived at the Antwerp central station it was time to check in at the Radisson Blu Astrid. I was greeted in the lobby by Lut De Baere (president) and Nadia Costantino of BOKS ngo (Belgian Association for children and Adults with a metabolic disease) BOKS ngo were the organisers of the conference.

The conference was designed with three target audiences – delegates, patients and families, and scientists and professionals. Each group had their own separate program in addition to a social common program, an industrial exhibition and a children's program.

Friday morning arrived and my attempt to arrive earlier to synchronise my body clock was a bit of a failure. I was up a bit earlier than expected. It was 3.30am and the communal breakfast at the hotel's restaurant 'Windows of Antwerp' did not start until 8am. Everyone started arriving by 7.30am and I quickly started to mingle.

The Friday morning session was opened by Lut De Baere – President of BOKS ngo and Nadia Costantino, followed by messages from Phillipe de Backer – Member of European Parliament, H Van Rompuy – President of the European Council and Laurette Onkelinx - Belgium Deputy Prime

Minister and Minister of Health and Social Affairs.

The biggest buzz would have to be the fact that we were in company of Royalty, HRH Prince Laurent of Belgium was in attendance as he is the Patron of BOKS.

Carmen from Queensland and I had a quick chat with him after the opening which was quite amazing as he took interest as to why two Australians were attending the conference in particular Carmen's situation as a PKU mother with a healthy little girl, all whilst being recorded by a local television crew.



Eric Lange – President of ESPKU and parent of two PKU children also took to the stage to welcome everyone. He was really cool and very funny as I was to discover later on the weekend. With the formalities over, it was now time for the information sessions to start.

First up, Professor Tref - a leader in Maternal PKU, highlighted the importance of maintaining a tight management of phenylalanine levels from pre-conception all the way through to delivery, in order to adequately protect the unborn babies especially from mental retardation and cardiac defects resulting from high levels of phenylalanine (Phe) levels and also growth deficiencies from inadequate protein supply during pregnancy. This topic was of extreme importance to Maria and I, Stella is now only 3 years old but there is no doubt PKU pregnancy is going to be a topic we will have to educate her on in the future.

Dr Schwartz followed with a talk on treating PKU in a very large country with few facilities. The next session was very interesting; an update of the development towards the European Guidelines for treating PKU was given by Prof. Van Spronsen. This process had been in conception for a number of years and it was making slow progress towards standardising the treatment throughout Europe.

There are many difficulties being encountered in this process, the main one being the fact that there are large differences between countries and centres in regards to treatment and management of PKU such as when to treat, what phenylalanine levels are optimal. Add 20 languages amongst the mix and you can see how some issues may be lost in translation. A consensus paper of patients with ideas about 'optimal care' in all European countries was yet to be published at the time of this meeting.

We stopped for a break. This was a chance to quickly visit the display area where Carmen and I met with the many sponsors that have come together for the weekend. We caught up with the VitaFlo team - what a great bunch!

A session in the afternoon that I also took interest was PKU @ School presented by Marlene Van Driessche. How to manage PKU while you are also trying to manage their education is very important for us as we strive to give our children a well balanced diet in order to give them every opportunity to perform at their best at school.

*In order for the children to succeed in their schooling years there is a need for information to be shared between all parties involved, an open channel of communication between your children's educators and yourselves needs to be kept at all times, you need to setup basic ground rules and there has to be a level of teamwork between the child and his/her family, the school and the metabolic team.*

Dealing with PKU at parties and holidays has not been an issue for us in the past, but Lucas will be attending the PKU camp in NSW later during the year so the next topic was also interesting. In Belgium there is a

*(Continued on page 11)*



# ESPKU Conference 2013 - A Fathers Perspective cont

(Continued from page 10)

not-for-profit organisation that runs PKU camps for children between the ages of 6-18 years. These camps are a fun escape for PKU children with the main focus being to educate about their disorder, share experiences and to 'connect' with others alike which will hopefully help them with the transition from childhood into adulthood.

All activities are well planned and a team of professionals including a metabolic dietician, social workers, supervisors, educators and resident chef is on site for the duration of the camp cooking PKU meals. A medical history for all campers including the protein tolerance is gathered and then nutritionally balanced meals are provided. The children learn the skills to manage their own diets by placing the onus on themselves in learning to become responsible and accountable for their own Phe-intake management, in an environment which has NO parents, only peers in the same situation.

*Xavier Duprez is a 30 year old Belgium national who gave us a very personal account of what it was like growing up with PKU and attending these camps. The many challenges that he has encountered whilst growing up brought both a smile and a tear to my eyes as I can see those same issues being mirrored on my Lucas. Needless to say Xavier has taken all that PKU has thrown at him head on, to make him the successful person that he is today.*

Following the Maternal PKU presentation by Marianne Diels, we met two ladies with PKU that became friends and eventually decided to start a family in their respective families. They told of their experiences through their pregnancies and how they eventually gave birth to perfectly healthy babies.

Although the conference had the main focus on PKU patients, it was attended by over 330 people directly or indirectly affected by over 80 types of IEMs from over 25 countries. One of those people is Joris who has tyrosinemia. He is now a healthy 9 year old who, on top of sharing an IEM with my son Lucas, also shares the same date of birth. I could not believe it when he told me this!

His mum told us about their experiences as an IEM patient living abroad, in particular Mozambique, where resources are very limited to say the least. Challenges in shopping for food and supplements, schooling and holidays were topics heavily discussed. They have to return back home in the Netherlands every 3 months to perform his plasma and liver tests. Living conditions are testing at times but that does not stop Joris from growing up and living his childhood like a child should.



**Joris and David**

Saturday morning was a very fun and interactive programme for all delegates, which included talks about Quality of Life (QoL) in day to day living with PKU, Micronutrients and Macronutrients in PKU and a presentation about IQ and PKU.



**Carmen, Nadia, Lut and David**

Interesting points raised were the relationship between early diagnosis and correct treatment with higher IQ levels in adulthood; the QoL and relationship between siblings with and without an IEM. The correlation between adult and childhood depression and anxiety was also addressed. There are no long term studies in regards to these topics which make it very hard to make conclusions in the long term. All information is cross sectional and age related.

Also, coming off diet in adulthood has been shown to cause abnormal complex executive functions, behavioural issues, psychiatric problems, depression, anxiety, cerebral problems, eczema and obesity. Therefore, the ESPKU guidelines (as we now do in Australia) recommend diet for life.

The final presentation was a questionnaire on PKU and treatment; it gave valuable insight into those who are directly affected by PKU and their opinions on the management of PKU. The conference concluded with speeches from professionals and delegates and a sneak preview of the country where the conference will be held in 2014, Croatia.

*We had some time to explore the city of Antwerp before the night's Gala dinner so with Nadia's husband as tour leader; we visited Antwerp Central Station, the museum of print and the Cathedral. At this point the ladies were in need of some well deserved shopping therapy whilst the males decided to explore a few watering holes.*

The Gala dinner gave everyone the opportunity to meet and greet further in a much more relaxed informal manner. It was a happy and sad occasion. It was happy because of all the friendships I had managed to create over the weekend; meeting all the delegates and sharing our experiences will be invaluable, sad because tonight it would all come to an end.

I briefly mentioned earlier the display area and sponsors that gathered here for the weekend. It was a real eye opener. The ease of access to low protein foods throughout the main countries in Europe really amazed me as well as the relative lower cost.

The VitaFlo team had a huge presence at the conference and throughout Europe; their team was very professional, helpful and knowledgeable. They made us feel welcomed throughout the weekend.

Another group that was present was Mevalia, part of the Dr Schar Medical Nutrition Group. Dr Schar is more commonly known for their provision of low gluten products for celiac

(Continued on page 12)



# ESPKU Conference 2013 - A Fathers Perspective cont

(Continued from page 11)

patients, but they have been busy researching and developing low protein foods for a number of years now.

The team from Mevalia introduced us to flours and pastas that they have developed. I met Nikos Charalampopoulos - Mevalia's business development manager. I briefly mentioned that I was Spanish and I was eager to find out if they had an outlet in Spain which he informed me that they did. I was visiting Spain following the conference so he put me in touch with Pablo Bazco - general manager for DR Schar for Spain and Portugal.

After numerous emails, I was invited to visit their Dr Schar office in Zaragoza, Spain. This was very exciting; I was going to visit the facilities where low protein foods were being manufactured. I could not wait!

I was picked up by Pablo himself and upon my arrival I was made very welcome as he proceeded to introduce me to integral members of his team. I met their head nutritionist - Neike and their marketing manager - Santi. I also briefly met their research team and consultants.

*Dr Schar manufactures foods in 8 locations around the world; each branch focuses on developing particular products. Research and development occurs in Italy where all core ingredients are manufactured with no additives or preservatives. From here they are shipped to the other branches where the different products are made and then distributed worldwide.*

The Spanish branch at Zaragoza focuses on making magdalenas (Spanish cupcakes). These are a staple food in the Spanish diet.

After putting on a white lab coat, Pablo showed me around the processing plant. First, ingredients stores and quality testing. Dr Schar has a very stringent process to ensure only the best ingredients are used. We progressed to the main processing plant where it all happens, all ingredients come together and the products are made. This conveyor belt style processing makes thousands of products per hour - all automated

24/7. From the mixing, to the pouring, to the baking, to the cooling, and finally, packaging. It was amazing to see it in operation and very overwhelming; it was one of those "you had to be there to believe it" moments.



**David visiting Dr Schar - pictured with Pablo (GM) and Naïke (Nutritionist)**

Before I made my trip I did some research to see what else was available in Spain to those patients that required a low protein diet. I came across a small family business Alecaran - operated by Dolores Blasco - mother of three children with LPI or Lysinuric (Lysinuric) Protein Intolerance. She was the worthy recipient of the Small Business Award in the category of Innovation at the local Business Awards in 2012.



**David with Dolores Blasco - operator of family business, Alecaran**

Dolores really struggled in the early stages with her first born. Continuously in and out of hospital for the first two years of his life, so much so they practically lived at the hospital. It has taken her over two years from concept stage, to set up her business which now makes low protein foods such as pizzas, sausages, waffles and vegetable lasagne. She also supplies other low protein products such as cheese.

I really look forward to keeping in touch and meeting up with Pablo and

Dolores next time I am in Spain. This trip has been an incredible and invaluable experience; it has been full of interesting and informative topics of discussion and speeches. The ESPKU 2013 in Antwerp has provided me, as a parent of PKU children, with the rare opportunity to find out how other countries are dealing with the diagnosis and treatment of PKU, learn about new low protein products available and spend time socialising with other families and patients with PKU from many countries.

I would like to thank my companion Carmen and her family for being there and sharing the journey with me.

Mostly I would like to thank **Robyn from VitaFlo** (sorry you could not be there). You have given me the opportunity to experience something that I had never thought possible. Thank you.

Thanks also go to the MDDA for supporting such an amazing venture.

*PKU is rare and often very challenging. We expend a lot of time and effort focussing on the day to day challenges of having to deal with such a complex disorder, that we sometimes forget to look ahead. It is nice to speak to people who have walked in your shoes and you look them in the eye and straight away they know and understand what you are talking about.*

I have found that the personal stories shared by people at the conference have made me realise that Lucas and Stella will be OK. That Ethan, Maria and I will be OK. We all will be OK!

David Contera



**MDDA would like to thank VitaFlo for their very generous sponsorship which enabled David Contera (WA) & Carmen Esparon (QLD) to attend the ESPKU 2013 conference held in Antwerp Belgium.**

## PKU Research at The Children's Hospital Westmead

Due to the efforts of Tiffany Dean (organiser of the PKU & Me Gala Ball), members and community we were able to donate over \$30,000 towards PKU research at The Children's Hospital at Westmead.

"I was delighted to receive the most generous donation from the MDDA in support of our PKU research efforts here at The Children's Hospital at Westmead."

"We are inching our way forward, but it is sometimes a frustrating process. We have been developing a genetically modified probiotic that we hope could break down dietary phenylalanine before it is absorbed into the bloodstream. Our initial studies were promising, but subsequent less successful studies are leading us to consider further modifications to our system."

"It is also very gratifying to see that several companies working in this space are interested in our progress and we are working towards developing a partnership with at least one of them so that we might be able to progress onto formal clinical trials in patients in the next couple of years".

"Your enthusiastic support helps bolster our efforts, and for this we are most grateful".

*Professor John Christodoulou AM*



## WA Fundraising Night

QUIZ NIGHT

Quizmaster John Lewis

Licensed bar and snacks  
available on the night

Great prizes to be won!

\$30 per ticket or \$200 for a table of 8

**HURRY!**

Pre sales only - no tickets sales at the door on the night

For tickets sales please contact

Scott	0413 169 664
Fiona	0419 935 918
David	0419 006 365
Maria	0414 185 911
Vanessa	0438 971 991

Saturday, 25<sup>th</sup> October 2014

7pm (doors open 6.30pm)

Bentley Community Centre  
Nyamup Way, Bentley

Supported by the Manning Rippers Football Club



Metabolic  
Dietary  
Disorders  
Association

ALL PROCEEDS RAISED GO TOWARDS THE  
METABOLIC DIETARY DISORDERS ASSOCIATION TO SUPPORT  
THOSE LIVING WITH RARE METABOLIC DISORDERS

[www.mdda.org.au](http://www.mdda.org.au)

## 2014 - 2015 Entertainment Books for PKU Research



All money raised will be going to Prof Christodoulou's PKU probiotic research.

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.

The books are available for different regions around Australia

Entertainment™ Memberships include over \$20,000 worth of valuable 2-for-1 and up to 50% offers from many of the best restaurants, arts, attractions, hotel accommodation, travel and much more!

Contact **Tracey Scott**  
[scott.tracey.a@edumail.vic.gov.au](mailto:scott.tracey.a@edumail.vic.gov.au)



# Introducing Kevin Alexander

Kevin Alexander is a PKU advocate and successful television news videographer living with PKU in Shreveport, Louisiana. Kevin was touched by numerous stories of patients off diet and sought to learn how he could make a difference to the lives of others living with this genetic condition. Here is Kevin's story...



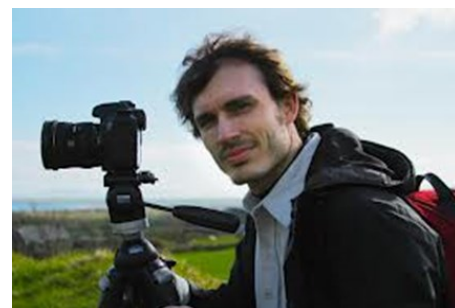
My name is Kevin Alexander, I have PKU, also known as Phenylketonuria. I'm currently in pre-production on a documentary web series called PKULife.tv.

PKULife.tv is going to be a series of short documentary films that profile individuals living with or otherwise affected by PKU. The seasons will be short, anywhere from 5-10 episodes per season. Season 1 will be shot in the US, and future seasons will be shot in various countries around the world.

I've been humbled by the response to My PKU Life, & through it I have discovered some incredible stories from around the world that simply need to be told. The best way we can continue to advocate for PKU, newborn screening or rare diseases is by sharing our stories.

As a creative professional and an adult living with PKU, I'm passionate about producing more documentary content for the PKU community. I've told my story but now I want to tell other people's stories.

Living with PKU can be a lonely experience. But we take comfort in



knowing we aren't the only ones living with this disease. Stories inspire, empower and unite. We may be scattered across the world but the boundaries that once separated us are dissolving. We can now be connected with one another no matter where in the world we live.

My goal is that these films continue to inspire, empower and unite all of us in the PKU community.

I am looking forward to coming to Australia to meet IEM individuals and families and to share your stories with the world.

Kevin Alexander, M.A.  
[www.pkulife.tv](http://www.pkulife.tv)

## Have your IEM stories filmed by Kevin Alexander

Kevin is looking to film interviews with individuals and parents of children diagnosed with IEMs here in Australia to share with the wider international IEM community. We would love to hear your experiences of living with an IEM and also caring for a child with an IEM.

### How will the Interviews be used?

Stories filmed by Kevin will be used as an educational tool for current and future individuals and parents of children with IEMs, healthcare providers and anyone interested in learning more about Inborn Errors of Metabolism

### How can I participate?

Filming will take place in Sydney prior to the NSW Family Retreat 2014 on Thursday 2<sup>nd</sup> and Friday 3<sup>rd</sup> of October.

If you are interested in being interviewed by Kevin and sharing your story we would like to hear from you - please email [office@mdda.org.au](mailto:office@mdda.org.au) or call on 1800 288 460 to arrange an interview time or to find out more information.



Thanks to the generous support of Nutricia, MDDA are bringing International PKU advocate and film maker Kevin Alexander to Australia for the NSW Family Retreat in October





## NSW Family Retreat 2014

**Friday 3rd to Sunday 5th October, 2014**

**Sydney Conference & Training Centre, Ingleside, New South Wales**

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.

The Family Retreat will be held immediately following the National PKU / IEM Youth Camp enabling youth campers to continue the fun and strengthen the friendships they will have forged at the camp.

### Some of the many activities to be enjoyed are:

- Social activities, fun team activities
- Speakers of Interest- including Kevin Alexander & Professor John Christodoulou
- Cooking Challenge - all involved
- Specialist breakout groups by transitional stage ie. kids/teens, adults, maternal, parents/families
- Special "Other/Rare IEMs Program"
- Clinic updates & latest research insights

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



## Major sponsor of the NSW Family Retreat 2014

MDDA would like to thank our generous sponsor VitaFlo for their substantial financial support for the NSW Family Retreat

## Grant opportunity - Tyrosinemia Type 1 families

The MDDA is pleased to announce we have been successful in obtaining a grant from Menarini Australia Pty Ltd to assist individuals and their families with hereditary Tyrosinemia Type 1 (HT1) to attend our 2014 Family Retreat taking place in Ingleside, NSW, October 3-5, 2014.



The grant will enable the MDDA to provide a Tyrosinemia specific stream at the event, and cover return economy airfares, retreat registration and accommodation for a number of HT1 individuals and their family/carer.

Please contact the office for an application form and guidelines at [office@mdda.org.au](mailto:office@mdda.org.au) or telephone Freecall 1800 288 460.

## Notice of Annual General Meeting

The Annual General Meeting of the Metabolic Dietary Disorders Association (ABN 99 021 391 381) will be held on **Saturday 4th October**, 6.00pm at **Sydney Conference & Training Centre, Ingleside NSW**.

The current Board of Management intend to renominate for their positions. Any members wishing to nominate for Board of Management positions may contact the office for a position description and nomination form to be returned by

**Monday 29th September 2014.**

Nominations for General Members are called for in all States and Territories



# Product Updates



Alpine Coconut Yogurt contains 1.9g protein per 100g,  
Available at Woolworths



Simply Wise Pizza Crust contains 1.5g protein per base,  
Available at Coles



Oreo Choc Wafer Fingers contain 0.7g protein per finger.  
Available at Woolworths



My Life Bio Cheese contains 0g protein per 100g,  
Available IGA VIC & NSW



Glutino Toaster Pastry contains 0.6g protein per serve



Organics 4 Kids Gluten Free Teething Rusk contains 0.3g protein per serve



cortexhealth

Cortex Health have now made available a range of great tasting and nutritious premium quality Low Protein Foods. Their range will be expanding regularly

To view their range and order online visit their website  
[www.cortexhealth.com.au](http://www.cortexhealth.com.au)

## New Loprofin products now available



Loprofin Dessert Mix  
(Vanilla & Chocolate)



Loprofin Animal Pasta



Loprofin Macaroni



Loprofin Chocolate Flakes

From August 1st 2014, all Loprofin orders less than \$100.00 will incur a delivery fee of \$15.00. Loprofin orders over \$100.00 will be delivered free of charge.

All customers who have ordered Loprofin products within the last 6 months will be notified directly by mail.

Nutricia Clinical Care Line 1800 060 051



## CONTACT US

**Address** Shop 4  
6 Thomas Brew Lane  
Croydon VIC 3136

**Postal** PO Box 33  
Montrose VIC 3765

**Phone** 03 9723 0600  
**Fax** 03 9723 0700  
**Freecall** 1800 288 460

**Executive Officer**  
Jenny Briant

**Email** [office@mdda.org.au](mailto:office@mdda.org.au)  
**Web** [www.mdda.org.au](http://www.mdda.org.au)

## COMMITTEE OF MANAGEMENT 2013—2014

**President**  
Monique Cooper (0414 734 061)  
(PKU Parent VIC)  
[President@mdda.org.au](mailto:President@mdda.org.au)

**Vice-President**  
Richard Drewitt (0429 075 131)  
(PKU Spouse VIC)  
[VicePresident@mdda.org.au](mailto:VicePresident@mdda.org.au)

**Treasurer**  
Paige Moore (0437 491 115)  
(PKU Parent VIC)  
[Treasurer@mdda.org.au](mailto:Treasurer@mdda.org.au)

**Secretary/Public Officer**  
Katy Drewitt (0488 777 241)  
(PKU Adult VIC)  
[Secretary@mdda.org.au](mailto:Secretary@mdda.org.au)

**Health & Wellbeing**  
Susi Hendricks (03 9592 8926)  
(PKU Parent VIC)  
[Wellness@mdda.org.au](mailto:Wellness@mdda.org.au)

## 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.

Email to: [office@mdda.org.au](mailto:office@mdda.org.au)

Photographs or hardcopies:  
PO Box 33 Montrose, Vic. 3765

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