



MDDA NEWS

Newsletter of Metabolic Dietary Disorders Association

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www.mdda.org.au

About the MDDA

The Metabolic Dietary Disorders Association is a national self-help group supporting people affected by genetic (inborn) errors of metabolism.

Our aim is to offer families a comprehensive resource of information and support. We provide members and their family's forums to share experiences and information, to educate themselves and the general community about living with an inborn metabolic dietary disorder.

The MDDA fosters co-operative relationships between members, healthcare professionals, government departments and other agencies to promote the health and well-being of members. MDDA objectives are pursued by the Committee of Management.

Reflecting on 2011

You can sometimes get caught up with the present without reflecting on the past achievements of the previous year. Well that's where we have been. With any new committee, members are keen to get involved and plan for the future. In this case the Executive Committee have been more focused on planning for the future than to provide updates of what has happened in the past with our members. We would like to thank all members for their contribution to the members survey in Dec 2011. These responses are guiding us into exciting new directions for 2012.

In this newsletter we are happy to provide an update about our recent *National Conference at Seaworld in Oct 2011*. This conference was a great success and I hope you enjoyed it as much as I did. We thank our sponsors for the event as without their support it would not have been possible. In providing further materials on sessions we have a 2 part article on *Maternal PKU—Planning for Pregnancy*. We then continue with an article on the *Adequacy of Medically Prescribed Diets: Does the Protein to Energy Ratio Matter?*

Our members have been active around the country with various gatherings and functions. It is great to see people get involved and to share their knowledge about the conditions they face every day. It is also a great way to meet new people and to also put a face to a name.

The articles on *Christmas in Victoria 2011* and *0-5yo Get-Together* are great examples of our members getting together. It's a great time for the kids to get together and play along with the

adults who were always keen to share stories and recipes.

Talking about recipes. We have added a *Easter Muffin* recipe this time around. Remember you can always check the website for further recipes.

The Doctors said this woman couldn't have children. This article was featured in the *Herald-Sun*. It is great story about what this Victorian mum has been able to achieve in the birth of her second child. Congratulations!

Our members in WA have done it again! The *WA Gala Ball 2011* raised \$10,000 for the MDDA. It looks like they had great night with lots of partying.

Each edition of the news is not possible without you sharing your articles, photos and news stories. We not only want you to share your thoughts and news but we also want to encourage our members to participate in the many activities that are planned. *Happy Reading!*

Richard Drewitt

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Disclaimer: Information presented in this newsletter is intended for general purposes only and should not be construed as advising on diagnosis or treatment of any medical condition, if you have interest in any of the foods or treatments contained in this newsletter check first with a qualified health professional.

Donations over \$2.00
are Tax Deductable



Easter Muffins

Adapted from Sharon Clarke
MDDA Cooking Demo 2000

Ingredients

225g Loprofin Low Protein Mix
½ tsp. baking powder
½ tsp. bicarbonate soda
½ tsp. cinnamon
½ tsp. mixed spice
54g sultanas *
2 tsp. Whole Egg replacer *
45ml vegetable oil
200ml water

Method

1. Heat Oven 180°C
2. Mix all dry ingredients together. Mix all wet ingredients together and add to dry ingredients to make a thick batter.
3. Place dessert spoon of mixture into paper cases or muffin tins and bake in a pre-heated oven for 15 minutes or until golden brown.
4. Set aside to cool completely.

Decoration

- ½ cup icing sugar
2 tsp. water
1. Place icing sugar in a small bowl. Gradually stir in water to make a thick icing. Place
 2. In a small plastic bag (snap locks work well). Cut one corner to make a small hole.
 3. Pipe a cross onto each muffin and set aside until icing is set.

Cooks Tips

- Cut in half and spread with Nuttalex.
- Cut in half and toast spread with Nuttalex and your favourite jam.

Note: Muffins freeze well.

*Calculate as guided by your dietician.

Enjoy!

MDDA National Conference 2011

The MDDA National Conference 2011 was held 15th-16th October 2011 at Sea World Resort—Gold Coast. With the theme "**Celebrating You**" it was a great opportunity to catch up with friends and make new ones. The event was a great success with close to 150 people participating across the two days.

- **Nutricia Masterchef Challenge** - what a creative bunch of chefs we had this year!
- **What's in my Pantry** sessions - thanks Louise for all the preparation and a stunning job facilitating this useful session.
- **Dr Jim McGill** provided such useful information on all IEM's (not leaving a stone unturned) and was eye opening for everyone in the room.
- **Bruce Lord** held a captivating session on the emotional side of living with a child with an IEM as we had the opportunity to explore the experiences and emotional phases of four PKU parents.
- Our **dietitian guest speakers Maureen Humphrey, Annabel Sweeney and Sue Thompson** provided useful insights into latest research, diet management and treatment options for both PKU and other IEM's.
- Our **International Guest speakers; Barbara Cochrane and Tricia Rutherford** who travelled such a distance to fill us with a world of knowledge from an international perspective and insight into even broader diet and formula options available.
- The **Teens breakout** session with **Mark Boyce, Rachel Sharman and Siobhan Brown** provided a great opportunity for our IEM teens to spend some time together sharing their experiences - and some great bonds were formed!
- Our **Maternal session** - whilst intimate in size - was rated extremely high in terms of the information provided by **Aiofe Smith** - Thanks Aiofe!
- And all of those that had the privilege of sitting through **Dean Tulloch's** presentation at the AGM evening dinner went on an amazing journey as he told his inspiring story of how he ran 250km through the Gobi Desert China all in the name of raising awareness for MDDA and for his little buddy Charlie (2 yo with PKU).



Maternal PKU: Planning for Pregnancy

Maternal PKU is the term used when a woman with PKU is pregnant. In this case it is necessary to maintain strict metabolic control both before and throughout the pregnancy to protect your baby.

Can a woman with PKU have children?

YES! Women with PKU may have healthy children if they maintain a strict diet prior to and throughout their pregnancy.

"I believe that the best thing that you can do is be prepared. There's always a risk that your baby may not be healthy. Every mother runs that risk and we have this as well as PKU considerations to deal with. But it is also possible, if we have very controlled diets to have very healthy, normal babies"

Why is it important to be on diet?

A high blood Phenylalanine (Phe) level in the intrauterine environment is toxic to the developing foetus. If the Phe levels are not controlled your baby may be at risk of congenital heart defects, slow growth, an abnormally small head (microcephaly), behavioural problems and developmental delay. Miscarriage may also occur. This is why women with PKU must plan their pregnancies carefully and maintain strict diet before pregnancy. For an unplanned pregnancy it is still important to return to and maintain the strict diet as soon as possible.

Why can these problems occur?

There is an approximate doubling of the Phe concentration across the placenta. The growing baby is therefore exposed to a higher concentration of Phe than that in the mother's blood. Phe levels that are safe for adults can be harmful to the developing foetus. For example if Mum's Phe level is 400 mmol/L then the Phe level her baby is exposed to is approximately 800 mmol/L. Any woman who has Phe levels over 360mmol at conception and regularly during pregnancy may be at risk of causing damage to her baby.

Is there anything I can do to prevent these effects?

The good news is that women with PKU who follow their diet to lower their Phe levels before and during pregnancy are more likely to have a healthy baby. Phe levels at conception and in early pregnancy are the most important to your baby's development; therefore it is very important to plan your pregnancy with your local metabolic centre.

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"It can be done and when diet is controlled chances of having defects are the same as anyone else"

" We get told all the bad stuff but we don't always hear that when we do the right thing and plan and adhere to the diet in pregnancy then we can have a healthy baby "

Is there any way to know if my baby will have problems related to maternal PKU?

A detailed ultrasound after 18 weeks of pregnancy can look for a heart defect or growth problem. Changes in intelligence, behaviour and facial features cannot, however, be determined before your baby is born.

"There are normally 2 scans during a pregnancy – one at 12 weeks and one at 18ish I think. The second scan shows the head circumference and this is the all-important one for us. You can elect to have a third scan, which I did – and which some doctors will request anyway, and this I did, simply for peace of mind. Many obstetricians may recommend this anyway as PKU is not something that may be amazingly familiar with."

What should you do when you would like to start planning Pregnancy?

It is important to contact your local metabolic centre for an appointment with your metabolic team as soon as you start to think about having a baby. Your pregnancy outcome is likely to be better where a supportive partner is involved in the counselling process- so please bring your partner along to clinic to meet the team. This will help your partner to understand what to expect and how they can support you along the way. It's a good idea to keep a food diary for a week prior to coming to clinic to give to your dietitian. If you have a protein counter try and estimate the amount of protein you are currently taking and write it in the food diary. Your dietitian will then be able to go through the food diary with you to check how you are counting protein, when you take your formula and the types of free foods eat. While in clinic ask your metabolic team about getting in contact with someone with PKU who has had a baby. This person may be able to act as a mentor for you during your pregnancy.

"Slow and steady wins the race.....It took a while for me to adjust to the low Phe intake. I found that slowly introducing less and less Phe in my diet helped me to commit to staying on diet rather than, as they say, going cold turkey."

"A good Support Network is vital."

Ok I have all the information I need to start planning what next?

Remember it's important to be honest with your metabolic team- they are there to support you. Let them know if you have not been taking any/all of your formula. They can help you access alternatives for you to try. You will need to take this consistently so it's important to choose the one you are most likely to be able to stick with. Let them know about any difficulties you are having with the diet or ordering low protein foods or getting bloods done. That way any issues can be ironed out early on making it easier for you to get your levels under control. Your team will probably request weekly bloods to assess current control. Clinics may vary regarding acceptable levels for pregnancy however ideal blood Phe levels for pregnancy are approximately 60 – 250µmols/L (<360µmols/L). Adequate tyrosine levels are also essential. Your team may request extra bloods to assess your status for other vitamins and minerals important in pregnancy. Aim to maintain your Phe levels in desired range for approximately 2-3 months pre-conception. Your clinic will advise you when your control is adequate and you are ready to try to conceive.

"An open communication network with your Specialist and Dietitian is important."

What can I do to help me lower my Phe levels?

It's important to order a range of low protein foods to help assist you with variety in the diet as your Phe exchanges will probably need to be reduced. You will need to complete the full prescribed quantity of formula every day and stick to the prescribed amount of Phe exchanges to achieve good metabolic control. Continue to have substantial meals if exchanges are reduced to avoid weight loss at this time. Remember it can take a considerable period of time to achieve levels in the desired range particularly if your Phe levels are high when you start planning.

" Be organised.....Plan meals including groceries and low pro foods, exercise, tasks/chores including baking and cooking into the week at the beginning of the week. It will assist in helping you stay in control and help you stay committed."

Aoife Elliott
Dietitian Team Leader
Paediatrics & Neonatology
Mater Health Services
Mater Children's Hospital. South Brisbane

Part 2 (I'm pregnant! What happens next?) will appear in next newsletter.



Adequacy of Medically Prescribed Diets: Does the Protein to Energy Ratio Matter?

Disorders of protein metabolism that require a restricted protein intake include individuals with urea cycle disorders and organic acidurias. While the goals of therapy in children include good metabolic control and normal growth and development, illness presents a period of high risk for these patients, and 'catabolism' or the breakdown of body tissue due to lack of energy must be avoided to prevent the accumulation of toxic metabolites.

When prescribing energy intakes for children with disorders of protein metabolism, the individual needs of each child must be carefully considered. However in general, requirements are based on values that have been determined for healthy children who are growing and developing well. Additional considerations for children with Inborn errors of metabolism (IEM) include extra energy to prevent catabolism, regimes to manage sick days when food intake is reduced, and extra intake for periods of high physical activity when energy needs are increased.

Similarly protein goals for children with these conditions are also based on requirements for well children and modified depending on how well the child is growing, if metabolic control is good and experience of clinical practice. In some instances the amount of protein prescribed may be pushed to the maximum amount tolerated to enable as free a diet as possible and allow for a more varied and 'normal' food intake. The protein: energy ratio (P:E) is a potential way to prescribe diets when both the energy and protein intakes are important, and in particular diets when the protein intake may be low.

The P:E ratio essentially is a way to describe dietary quality by asking the question "If an individual or group consumes this diet, in amounts that will satisfy energy needs,

will the concentration (density) of protein also be high enough to meet protein needs?"

In the past P:E ratios have been used to determine the safety of diets in developing countries, and they have also been used in recommendations for the protein content of infant formulas and for single food diets such as specialised feeds. They can also be used when a group of people is eating the same diet such as in a nursing home or hospital.

There is also the potential to use a P:E ratio recommendation for those requiring a very low protein diet such as those with disorders of protein metabolism. P:E ratios could be used to assure the diet has:

- adequate quality, particularly as food sources of protein can contain an important supply of other nutrients such as iron and calcium.
- the protein in the diet needs to be of sufficient quantity to ensure growth and so that the body uses protein most efficiently, and
- to ensure that the diet does not become overly diluted by the use of excessive carbohydrate and fat.

In order to do this, more information will be required about the protein and energy intake of patients with IEM, how often their protein intake needs to be reduced to manage sick days, their nutritional status, their body composition and their energy needs. This research into the longer term effects of low protein diets has commenced at the Metabolic Service in Melbourne with the hope that this will help provide more information about the most effective ways to keep our patients well and healthy.

Maureen Humphrey AdvAPD
Clinical Specialist Dietitian.
Metabolic Genetics
Genetic Health Services Victoria
Royal Children's Hospital Melbourne

Thank You for the following suppliers that supported the conference:



MDDA Matters

AUSTRALIA DAY HONOUR

Debbie Colyer has been awarded an Order of Australia Medal for her work in educating and supporting others living with PKU. On behalf of our members in NSW, MDDA congratulates Debbie on her outstanding contribution in her work with the PKU Association of NSW.

IT'S A GIRL!

Anna Hoar (PKU WA) had a beautiful baby girl, Emily Kathryn on 20th February 2012, weight 3.75kg and 51cm long. Both Anna and Emily are doing well.

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. Any questions please contact the Department directly on (02) 6289 8980. Or contact us at the office.



Rare Disease Day was on 29th Feb 2012. What did you do? Let us know?

Christmas in Victoria 2011

On a very windy Sunday afternoon at Albert Park Lake in Melbourne, ten families got together to catch up and celebrate another year gone by. With everyone generously bringing a plate for all to share there was plenty of yummy low protein tasters to try: Cake, Carrot muffins, fruit kebabs, pasta salad, antipasto platters, chocolate cup cakes, jellies, popcorn & lollipops ensured no-one went home hungry.



The ages of those with PKU ranged from adult right down to gorgeous little Harrison at 8 weeks so lots of recipe ideas, lifestyle factors, and ideas on how best to live with a special diet were shared. A few laughs were had on the perspective of the 'grown ups' with PKU and how they dealt with their food at their young age. For me, as a mum of an 11 year old with PKU it was re-assuring that I'm not the only one whose dishes don't always turn out as we thought they would, or even if they do - they're not necessarily 'liked'. Despite all the trials and tribulations we all marvelled at how we adjust, kids adapt, and most importantly we have gorgeously healthy, full of life, little people (and not so little people).

The kids had plenty of energy to enjoy the playground and a late afternoon cricket match and it was great to see them all have fun and enjoy the day. Luckily the weather held on and despite some threatening rain we all managed to stay dry. Gotta love Melbourne - even in December we can't always be sure we'll get a rain free day!

Susi Hendricks

Doctors said this woman couldn't have children

Leah Cattapan is believed to be the first woman in the world with two rare metabolic disorders to have not just one, but two children. When her eldest daughter, Lara, was born two years ago, she became one of just a handful of women worldwide with her condition to bear a child. But a determined Mrs Cattapan has stepped it up, giving birth to her second daughter, Ashley, a fortnight ago at Monash Medical Centre, making medical history.

Doctors had advised her against having children because her genetic metabolic condition was too risky, as she could die during pregnancy or soon after birth. The hospital's adult inherited metabolic

disorders service head, Dr Gisela Wilcox, said Mrs Cattapan was unable to break down protein properly.

When she metabolises protein she produces dangerously high levels of ammonia that are toxic to her brain and can cause confusion and, left untreated, a coma and death. Mrs Cattapan, 35, also suffered a related genetic condition that left her prone to infection, Dr Wilcox said.

She took her precious bundle home yesterday, after spending a month in hospital for the birth, where her blood ammonia levels and nutritional intake were closely monitored.

A generation ago it was out of the question for women with these illnesses to have children, but medical advances had now made this possible, Dr Wilcox said.

Mrs Cattapan had a 50 per cent chance of passing her illnesses on to her children, but neither Lara nor Ashley suffered from or carried either condition. "She just wants to get on with life, and be a mum ... to be a parent is very meaningful for her," Dr Wilcox said.

The Blairgowrie mother said she was overjoyed that neither of her children were affected by her illness. "They are very precious to me, and I thank God every day that they are healthy, and they haven't got what I've got," she said.

Marianne Betts

bettsma@heraldsun.com.au
Source: Herald Sun 22/12/2011



Picture: Mike Keating Source: Herald Sun



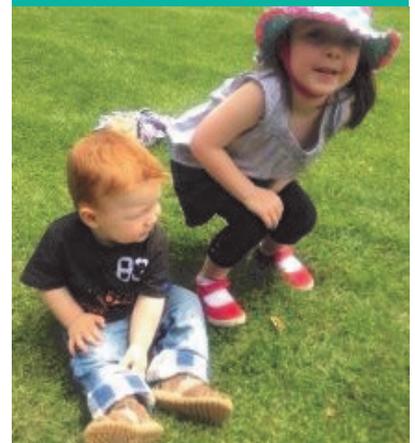
0-5yo Get-Together

Our 0-5yo Get-Together in November was a wonderful sunny day – perfect for a visit to Chesterfield Farm—Victoria! Myself and son Reef (PKU), along with Tiffany Dean and her two children Matilda and Jesse (PKU), and Tracy Scott with new baby Harrison (PKU) had a lovely day at the busy farm, Matilda even lucky enough to milk a cow! It was nice to see the children playing together and as always, wonderful to



exchange recipe ideas and stories. Tiffany and myself were able to tell Tracey about our boys' journeys with PKU so far, and try to help her feel a little at ease with baby Harrison's recent diagnosis. Harrison was a perfect angel coping perfectly with the screams of the school groups around us! Though there were only a small group of us we all got a lot out of the day together – just having people who 'understand' the day-to-day issues PKU brings, and we all look forward to coming Get-Togethers. In no time all 3 of the boys will be running around together.

Wendi Emmerson





WA Gala Ball 2011

Last year Cleopatra's Temple Beauty and Day Spa organized an extravaganza of a night – a black tie ball with the proceeds being donated to the MDDA. It was an amazing night, with entertainment that included a battle dance by gladiators, fire throwers, Italian singers and a great band to dance the night away to.

Cleopatra's Temple Beauty and Day Spa is located in Perth. The owners of the Spa are Rocco and Claudia Mazzone, brother and sister-in-law of Maria Contera and Frank Mazzone who are members of the MDDA.

The ball was held in the Grand Ball Room of the Duxton Hotel in Perth. Anyone who has visited the hotel will know that this made an elegant and grand backdrop for the night. Pre-dinner drinks were served in the foyer of the hotel. Roman gladiators flanked both sides of the entry to the ball room when guests were ushered in. The ballroom was



Claudia and Rocco Mazzone
L-R Lucas Contera (PKU) Stella Contera (PKU) and
Nicholas Mazzone (PKU)

decorated beautifully and the scene provided a feeling of grandeur and drama, which was further accentuated by the theatrical battle dance of the gladiators and fire throwing display.

Kaye Rabbone played the part of Cleopatra, and was a fabulous MC for the night. Her entry was on a Roman bed, held high by Gladiators. The meal began with a choice of entrees. I chose the Ravioli and this was superb, one of the best dishes I've ever tasted. The entire three-course meal was fabulous, and we were entertained with Italian singers who moved throughout the room during the evening.



The band Retro provided great music to dance to, demonstrated by the many people who ventured to the dance floor and 'boogied'. Rocco Mazzone certainly proved his talents as a singer! The competitions during the night, for which there were assorted prizes, added to the fun of the night, as well as the silent auction for a large number of assorted items.

What a night it was. The event can be succinctly described as 'spectacular'. Congratulations to all involved in organising the event, particularly Rocco and Claudia Mazzone and Cleopatra's Temple. Thank you also for naming the MDDA as beneficiaries of the proceeds of the ball, as they did the previous year. They sure know how to put on a party!

Shona Browne

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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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