MDDA Matters

Issue #74 - December 2021

Metabolic Dietary Disorders Association

Merry Christmas and Happy New Year!

As the end of 2021 approaches the MDDA reflect on a year that once again brought about new challenges.

We have been able to maintain connections with our community through our Virtual events Member Connect Hub and also as the host country for the Global PKU Patient Conference that was held in September. These events have all provided a great opportunity for our community to come together to connect, share and learn.

MDDA would like to take this opportunity to thank everyone who has contributed in any way this year. We are extremely appreciative of all of our members who have taken part in sharing their stories - whether that be through submitting their story in our newsletters, joining us online in our Member Connect Hub - Spotlight series, or making a telehealth call to our Pathways nurse & sharing your patient IEM journey to contribute to rare disease research. We know it's not always easy and time is precious, so we want you to know how much you are appreciated and how you have helped others just by sharing and letting then know they are not alone. The MDDA relies on the support and dedication from our members to further our initiatives and educational programs. Thank you!

MDDA continue to be effective, productive and valuable in giving a voice to the IEM community. Our Executive committee consistently lead us in education, connecting, support and advocacy, working toward MDDA's mission, vision and goal. We are extremely lucky to have such an active committee and thank our amazing volunteer executive committee, Monique, Louise, Paige and Bianca.

In our last edition for 2021, we once again share with you inspiring stories and updates on things happening in our MDDA and IEM space. If you would like to participate in our Hub or contribute to our newsletter in the future, please email the office, we would love to hear from you.

We wish everyone a very Merry Christmas and a safe, healthy and happy New Year! We look forward to sharing 2022 with you all.

in this issue

MDDA Office Christmas closure:
The MDDA office is taking a break from the 22 December to 10 January 2022.
For the remainder of January our office hours will be reduced.

MDDA Member Connect Hub

As mentioned in our last Newsletter, in June we launched the **MEMBER CONNECT HUB**. The Hub is a way to engage the virtual space to help keep our members engaged and connected during a time where we still can't meet in person. An inclusive and interactive virtual platform, the Hub is a platform for MDDA members to connect and meet with other members, gather in special interest groups, forums and attend virtual events and workshops. We have been running sessions that have been a great mix of informative and social sessions that have engaged the entire IEM community. Make sure that you keep an eye out on our social media to find out the Hub sessions we have planned for the rest of the year and into 2022! Email connecthub@mdda.org.au for your login.

MEMBER CONNECT HUB

If you want to find out more about the Member Connect Hub please visit mdda.org.au/hub

Session Features

ADULTS AND PARENTS

ED CONNECT FORUM

Educational webinar style presentations

ADULTS AND PARENTS

SPOTLIGHT SERIES

Intimate 1on1 or 'on the couch' panel style member interviews with audience interaction

ADULTS AND PARENTS

VIRTUAL HAPPY HOUR

Casual Virtual Networking through the Member Connect Lounge and various Chat Rooms and our Connect 4 Speed Networking

KIDS AND TEENS

CONNECT GROUPS

PeeKabU Kids Mentoring Groups with Low Pro Teens (monthly challenges & show & tell)

KIDS AND TEENS

TEENS LEADERSHIP WORKSHOPS

Our Low Pro teens leadership team get together for coaching and guidance to support their IEM management and guide our PeeKabU kids with mentoring skills

KIDS AND TEENS

TEENS PANELS

Designed for Parents and other teens to come and ask questions of various IEM Teens

Thank you to our Hub Sponsors!

Platinum Hub Sponsor

NUTRICIA
LIFE-TRANSFORMING NUTRITION

Gold Hub Sponsors

Vitaflo
Enhancing Lives Together
A Nestlé Health Science Company

PTC THERAPEUTICS

Previous Spotlight Sessions

The Spotlight series are intimate one-on-one panel style member interviews with interactive audience interaction. Over the last few months we have had the privilege to hear from our very own inspiring IEM community. We thank each of them for allowing us to delve into their lives and hear about their life journey, experiences and challenges. These honest reflections are available to view in our Member Connect Hub.

Debbie| PKU Adult
A wonderful way to start our spotlight series was with Debbie and mum, Jill Colyer. They provided a look into the history of PKU and the journey of diagnosis and treatments in Australia over the past 50 years.

Daphne Doukas | MMA Mother
A conversation between MDDA Vice President Louise Healy and Methylmalonic Aciduria mother and long-time MDDA Member, Daphne Doukas, provided an insight into the challenges faced and her advocacy role .

Anita Inwood | Director of QLD Lifespan Metabolic Medicine Service
Anita led the informative presentation and discussion on the new Maternal Women National guidelines for Sapropterin.

Chelsea Docherty | PKU Adult
We heard from Chelsea, whose story showed there are no limits on achieving. She is currently completing a masters degree, working in Cyber Security and competes in the sport of crossfit .

Frances Del Castillo | PKU Teen
Frances and her Mum, Beatriz, shared with us how she managed her school years, part-time job, social outings and transition to university.

Brooke Hall | OTC Deficiency
Brooke took us on a journey in living with OTC Deficiency. We heard how this positive 24 year old is fitting in studying to be an Occupational Therapist, while working and enjoying all that life offers.

Wilson Family | PKU Adult and Baby
The Wilson Family family joined us from Tasmania and told their story of both Mum, Morgan and baby, Annabelle who both have PKU. It was also great to hear from Ben, who provided an insight from a Partner and Dads point of view.

Tammie Rees | Mother of Ava MSUD
Tammie told their story of how Ava was diagnosed at 16 days old with Maple syrup urine disease, and the positive impacts of her having a connection with an older MSUD teen.

We have had some wonderful encouraging feedback in response to our Member Connect Hub. With each session we have seen growth in members connecting in for the first time and more return users to the hub with each new session.

Our live cooking session with Jude from Vitaflo proved popular with new parents and grandparents. It was also great to have our young members jump on line and be involved in our Teens panel. This new initiative brings our community together, enabling personal connections during these times where our members can't gather together. We are learning as we go, supporting one another as we navigate new ways of connecting, supporting and educating – providing new ways that we can best connect with our members and families.

gap GLOBAL ASSOCIATION FOR PKU
Bridging the gap for PKU worldwide

Around the world in a day Virtual event experience

MDDA were delighted to be the host patient group and organiser for the Global Association of PKU's (GAP) inaugural Global PKU Patient Conference that took place online from the 10th to 11th September 2021.

The first of its kind, this event brought the PKU patient community together to celebrate our achievements, learn about new developments and move one step closer to advancing PKU as a global health priority, accelerating new knowledge, treatments and a cure.

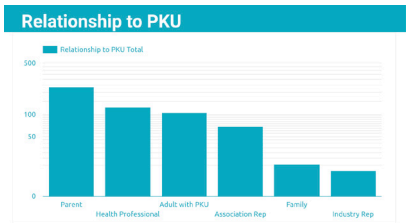
The conference was a showcase of PKU scientific endeavours including research, studies, clinical trials and best practice care for PKU patients on a global scale, informing and inspiring attendees on what the future has to hold.

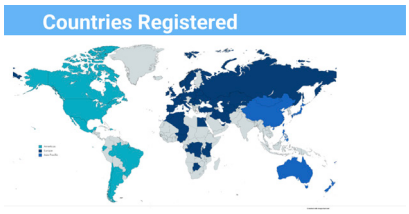
PKU patients and the wider community, health professionals and the scientific community from over 69 countries came together for an informative and inspiring interactive virtual event.

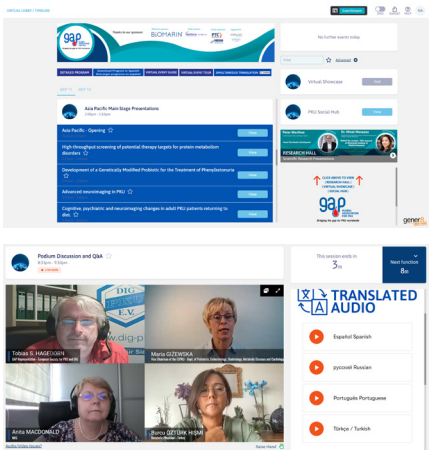
The event took part over a 24 hour period being hosted in 3 separate regions of the globe including Asia Pacific, Europe and the Americas. The live presentations featured simultaneous live translation in addition to live Panel Q&A discussions, Chatrooms, scientific showcase and the opportunity to chat 1on1 with scientists and researchers.

Australia was proudly represented with live interactive presentations from Prof John Christodoulou, A/Prof Gerard de Jong, Prof Mark Walterfang, Dr Michel Tchan, Dr Minal Menezes and Catherine Manolikos (Dietitian).

Thank you to all the people who attended and made this event something unique and special as we came together as a global PKU community!







Platinum Sponsor
BIOMARIN

Gold Sponsor
NUTRICIA METABOLICS

Silver Sponsors
PTC THERAPEUTICS
Nestlé HealthScience

Supporters
synlogic
HOMOLOGY Medicines, Inc.

Patient Pathways Program
Meet Jo Campbell

As a Patient Pathways Nurse I am here to support MDDA members, this is a FREE telephone support program.

My role is to provide a central point of communication and engagement to ensure optimal care and coordination of services along a patient's entire health care journey. I am available to assist you to put in action the care plan you have received from your metabolic team and general practitioner.

I am available to educate and help patients to better understand their health conditions and enable them to self-manage, participate in decisions about their health care (eg, clinical trials) and improve their own health outcomes.

I enjoy working with the MDDA team and providing input to our varied programs and resources eg Zest, including collaborating with the multiple MDDA stakeholders.

PKU PATHWAYS

Each week I speak with a number of new and existing members. I welcome our new members onboard, outlining how MDDA can support living with an IEM and helping to direct them to answers to their questions. Some of the common areas I help with include, finding about access to services that you may be eligible for, informing members of different health avenues, assisting members to prepare for upcoming GP and medical specialists consultations.

I am passionate about educating and supporting members throughout their challenges and I have been able to work with so many delightful MDDA families and individuals.

My service is available to ALL members of the IEM community, no matter your age, if you have been with us for a number of years or recently joined. Don't hesitate to call me, we know different health questions arise at different times.

If I haven't connected with you please give me a call or email, and lets arrange a time to chat! I would love to get to know you and see if I can be of any assistance in helping you to manage your or your families IEM.

03 9723 0700
pathwaysnurse@mdda.org.au

Advocacy Update

Louise Healy, MDDA Vice President, has continued her Advocacy role with the MDDA. We thank Louise for representing MDDA and continuing to provide a strong voice to government and other bodies. Below outlines MDDA advocacy plans for 2022.

The 3 big advocacy priorities for MDDA are;

- Access to treatment
- Contributing to policy change
- Clinical care and resources

The focus of MDDA advocacy continues to be equitable access to treatment and care, and over the coming year this will include clinical trials, in particular for the rarer IEMs and advocacy around reimbursement for treatments that have been approved as safe and effective. The focus on policy change will continue to look at ways that the system can more equitably assess cost effectiveness and evidence requirements for rare disease therapies as these have been some of the main barriers to getting access to treatment.

In the new year, we plan a series of education sessions so that people can better understand the regulatory and reimbursement process in Australia - this is intended to support individuals who wish to do any advocacy to make it is as effective and targeted as possible.

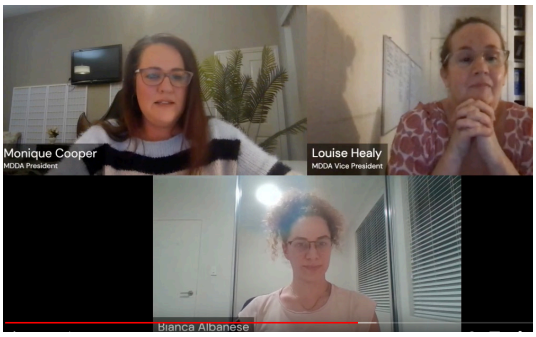
As part of ongoing broad advocacy MDDA will launch an awareness campaign highlighting the challenges of managing a lifelong IEM.

How you can help

- attend our online events, meetings for regular updates.
- reach out to your local federal MP and make them aware of PKU, your personal situation, the inequity around access to sapropterin for adults and the public comments made by the PBAC.
- Adults can provide a written statement to add to the evidence we are compiling.
- Participate in research – surveys

In the coming year our plan includes

- Advocating directly to companies regarding reimbursement submissions and clinical trials
- Ongoing engagement with PBAC
- Engage with political representatives
- Public awareness campaign – continuation of video series
- Continue to write submissions - to raise barriers
- Continue working with ASIEM
- Launch contact registry
- Continue to collect data and evidence – we will encourage you to participate to help
- **Call to action – get involved!**



MDDA AGM

MDDA held our virtual AGM on the 9th November and welcomed back into office all our Executive Team from the previous year, Monique Cooper - President, Louise Healy - Vice President, Paige Moore - Treasurer & Bianca Albanese -Secretary. We would like to thank them all for their incredible leadership and support this year.

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

MDDA are extremely lucky to have this commitment from the executive team.

Along with the yearly reporting from our President, Vice President and Treasurer we also heard from our Patient Pathways nurse, Jo, Executive Officer, Jenny and Bella from our communications team who also provided updates in their areas. A recording of the AGM is available in the Member Hub or on our YouTube channel.

Thank you to all of those members who attended. Our annual report will be available online early December through our website mdda.org.au. Should you wish to receive a hard copy please contact the office.

Lachlan's Story

Thank you for the opportunity to share our story which I know many of you will identify in some way with your own experiences.

If I am completely honest the years have been nothing short of an emotional rollercoaster. Turmoil, constant worry and fear of the unknown mixed with the joy of reaching a milestone, gratitude for each day, each birthday, never taking life for granted.

Lachlan was born 2 November 2002 and diagnosed with MMA Mut-0 by the newborn screening test at 12 days old. He was admitted into the RCH, ICU in metabolic crisis. Like many of our warriors, Lachlan's start in life was rough. Although his birth went well, initially he had slight jaundice and wasn't feeding properly but nothing to worry about (or so we thought). We were excited to be given the green light to go home and start our new life as a family. Over the next couple of days things didn't feel quite right. We arranged a lactation nurse to visit to help with the feeding issues and mentioned the groaning noises he was making during his night sleep.

Concerned, I rang the paediatrician who put my mind at ease by reassuring me we weren't experiencing anything different from any new parents settling in and if we walked through the hospital nursery at night time, we would be surprised at how noisy babies could be. But the following day I noticed Lachlan had vomited some of his feed while he was sleeping. I questioned whether this was normal and although my gut feeling was growing concern there were no other obvious signs.

Our saving grace call came that afternoon when a doctor from the Royal Children's Hospital rang to say Lachlan's newborn screening test had shown a high level of chemical in his blood and to bring him into hospital immediately.

Little did we know at the time "the doctor" was Head of the Metabolic Team. From the moment we arrived at the hospital, while the medical team appeared calm things moved fast. Lachlan was admitted and as the hours passed Lachlan's condition deteriorated. "Dr. B" took my husband and I into a little room nearby to tell us they had established our son had a rare metabolic condition called MMA, possibly Mut-0, as he was non b12 responsive. He was in metabolic crisis and they were transferring him to ICU.

While they were doing everything possible, the reality was he may not survive and suggested that if we wanted to call in family then not to delay. We were in our worst nightmare. I remember sitting in that little room in ICU with my beautiful (now late) husband, both of us in complete shock praying to God to just let him survive and we would take him any way he came and do everything to make his life as best we could. As immediate family arrived "Dr B" came in again and somehow very calmly explained the process of treatment and said the next 24 – 48 hours were critical.

Although the chain of events of those next 48 hours are somewhat of a blur, some of those images and feelings are still so clear to this day. Miraculously, thanks to the incredible work by "Dr B" and the metabolic team and the person in newborn screening who picked up the discrepancy in the first instance, our little fighter survived. After a number of weeks in the neo natal ward, our little "chipmunk" was coming home. I called him

this with love as when we were finally able to leave the ward for a walk within the hospital there was this tiny little baby in a big hospital pram with his great big beautiful brown eyes peering over the blanket.

Taking Lachlan home was both incredibly joyous and daunting as we had so much to learn.

Our heads were in a whirl calculating daily intake of protein, calories and medications not to mention being first time parents, but we didn't care as we felt so incredibly lucky. For years, each time we visited the hospital for Lachlan's clinic appointments, the amazing "Dr B" (who was Head of his metabolic team for next 16 years) would always say to Lachlan "here is the young man who caused me some sleepless nights".

Fast forward almost 19 years, with countless hospital stays to keep him stable due to gastro or other childhood illnesses, doctors and therapists appointments, blood tests, nasal gastric tubes and at the age of 4, the devastating loss of his father under his belt, we have the most remarkable young man in our lives. Yes, he has his health challenges, his weekly medication box is pretty full and he is closely monitored by both the renal and metabolic teams but

Lachlan's positive outlook on life, his tenacity and his resolve, his strong will not to let MMA define him gives me the strength I need. To anyone who ever says you can't - he says I CAN.

Despite all the bumps along the way, Lachlan successfully completed Year 12 VCAL and is currently doing a furniture and cabinet making course which he loves. He works part time, is learning to drive and can't wait to buy his first car.

Whilst we always tread with an element of sensible caution, we have also done our best to ensure Lachlan participated in activities others wouldn't think twice about.

Like the time in secondary school when he was desperate to attend a four-day camp to a remote part of bushland in Victoria (Australia) with no mobile reception. Both sides of my brain started their usual battle as they had done so many times before like when he was 3 years old trying to decide if I should send him to day care "with all the germs" until his metabolic doctor suggested quite matter of factly "the sooner he goes the better, as you can't wrap him up in cotton wool." Here we go again - how could I entrust someone else with his care in this environment? How would we get him out of there quickly if he became unwell? But we went to work.

His wonderful outdoor education teacher was so happy and went to great lengths placing extra vehicles along the way (although he did say he would carry him out of there if need be), accommodating his food requirements and medical management plan with the promise he had the satellite phone and yes he assured me the battery was charged.

To ease my mind, I also contacted Air Ambulance Victoria to ask about their process and length of time it could take to get someone out of the remote bush in the case of an emergency. They were incredible and offered to work with the teacher to map out exactly where they were situated and entered all of Lachlan's medical emergency plan into their system so that in the event, they received a call they were ready to go. Lachlan had an amazing four days and arrived home unscathed, tired but elated to have had this experience with his school mates. We all slept well that night.

Obviously the MMA journey affects everyone in the family and I am not going to lie it has been a tough gig at times particularly as a sole parent juggling full time work and the mental load of the constant underlying worry but we have muddled our way through with the help of supportive employers & work colleagues, family and close friends who I have been able to reach out to when three wheels have fallen off the bus and the fourth is about to go, who drop everything to help in an instant without question or judgement.

Although it's hard to ask sometimes, I have learned people want to help. These people I cherish and am indebted to.

Lachlan also has a wonderful 15-year-old brother Christian (unaffected) and the bond between them is very strong. Over the years, Christian has been ferried off to family or friends during hospital stays, sometimes in the middle of the night, last minute cancellation of parties, weddings and holidays when things took a turn, accepting that's just the way it is without complaint. I am so extremely proud of Christian being so caring and supportive of his brother and I know it is not easy on him as he worries about his brother too.

They are both incredibly resilient young men and that is I guess what adversity creates. This journey has given us the awareness to cherish our children and embrace life in a way that perhaps we may not have otherwise.

Although it's hard to stay in the present and not think too far ahead given the progressive nature of MMA, I try to reflect on my late husband Victor's words to me, "don't worry about what tomorrow will bring, just do today". We have so much gratitude for the incredible Metabolic Team at The Royal Children's Hospital and now The Royal Melbourne Hospital where Lachlan has transitioned as an adult. We are now riding on the wings of hope that one of these new treatments will come sooner rather than later, work and work well as without hope we have nothing. Blessings to you all – I feel we are an extended family, understanding each other's journeys, in ways others cannot, differ as they may, and I am thankful. *And to Lachlan and Christian if you read this, you are my superhero warriors.*

Daphne Doukas
MMA – Mut 0 Parent, Melbourne Australia

My role with the MDDA is the MMA Link representative, in the coming months we hope to work with families with other organic acidemias to leverage common issues, especially with some emerging clinical trials targeting more than one of the OA conditions. Our mission is to continue working towards raising awareness and a key advocacy priority to work with companies who have clinical trials to establish Australia as a trial site.

A high priority for the MDDA is the establish a contact registry including capturing all rare inborn errors of protein metabolism. The purpose of this registry is to provide data to organisations considering research and or clinical trials in Australia and position Australia to take advantage of emerging therapies, like gene and cell therapies that offer hope to families for potential treatments. Australia is fortunate to have very experienced and dedicated clinicians who have expressed interest and are very supportive of our efforts for this to happen. We hope some of the gene therapies that are emerging will ultimately be of benefit to many of the rare protein IEMs. I can be contacted at daphne.doukas@mdda.org.au

In Memory of Koby

MDDA and the IEM community are devastated and heartbroken by the tragic loss of our much loved and admired member of our community Koby and two of her beautiful children Chloe 8yrs and Harrison 20 months. Our deepest sympathies extend to her loving partner Craig, and their two sons - Lachlan aged 9, and Liam aged 6.

Koby was a very active member of our IEM community always supporting others and playing an important role in advocating for the needs of patients living with Tyrosinaemia Type 1 – a rare metabolic disorder that their two boys Lachlan and Liam live with. Koby was a generous, strong, humorous, down-to-earth and caring woman. Her passion to support and advocate for her boys and the wider IEM community was an inspiration to the MDDA and those of us who had the privilege to know Koby.

Koby you were one of a kind and will be deeply missed.

A gofundme page has been created to support Craig and the boys during this most difficult time for them:
www.gofundme.com/f/in-loving-memory-of-koby-chloe-and-harrison

Sophia's Grade 5 Camp

In January this year our family moved from Ringwood East, Melbourne, Victoria to Airlie Beach in Far North Queensland. Sophia, my 11yo daughter with PKU began grade 5 at St Catherine's Catholic School in Proserpine.

The week before school started we had the opportunity to go to school and meet the classroom teachers and go through Sophia's PKU management. Can you believe there is another kid at the school with PKU and not only that, he is also in Grade 5 – Kohen.

Kohen has been going to St Caths since he was in Prep and his older sister in the senior school also has PKU. So not only was the school very educated and aware of the PKU diet and requirements , but Sophia now has a classmate with PKU.

In term 3 our school was fortunate enough to be able to go on school camp. The camp was about 120KMs from home for 3 days and 2 nights at Kinchant Dam. Most parents of kids with IEMS know this can be a very anxious and daunting time as there is a lot to prepare for camps. *The camp operators were fantastic and supplied the full menu ahead of time in an excel spreadsheet.* I was able to colour code the spreadsheet to mark up Okay/ Not okay and in moderation items and I then supplied the alternatives. (Breads, pastas, snacks, milk, etc).

Natalie, Kohens mum also went on camp and was great in helping Sophia, camp staff and educators with the diet and making sure she had her formula. Some of the items on the menu I needed to be creative with to come up with similar alternatives like hot dogs, sausages, crumbed chicken, bacon and eggs.

Soph is on about 7grams of protein, so there are not many store bought alternatives. I was also trying to make or use snacks that did not need weighing or a lot of preparation. I marked up the spreadsheet with the quantity for the 'in' moderation foods. For example, Arnotts sweet biscuits x 2. Below is my alternative list for foods that were Not okay items. Also, many of the snacks on camp were suitable for Sophia such as fruit and veggies.

Breakfast Alternatives	
Toast	Crumpet – Liberate Crumpets 0.5g each
Bread	Home made bread - Loprofin
Bacon & Eggs	Mushrooms and Tomatoes Asparagus
Milk	PRO ZERO MILK
Wrap	Genius Wrap
Cereal	1 cup of Rice bubbles -
Lunch Alternative	
Sliced Cheese	Bio- Cheese slices
Flat Role	Genius roll
Bread	Home made bread - Loprofin
Long Roll	Genius roll
Weiners	Not Dogs
Grated Cheese	Grated Bio Cheese
Dinner Alternatives	
Patties & Sausages	Baked Mini-Frittatas With Veggies & Cheese https://www.cambrooke.com/recipes/low-protein/recipe-detail.php?id=1469#.YXvBQJ5ByUk
Sliced Cheese	Bio- Cheese slices
Flat Role	Genius roll
Bread	Home made bread
Crumbed Chicken	Cambrooke Chicken Patty's _ Pre-made

Something else I did was prepare the food and snacks with Sophia, that way she knew what she could have and when she could have it. It was also fun for her as she enjoys cooking, and she could have a lot more fun on camp, doing activities and not be worried about food and being hungry.

On the morning of dropping off Soph to school for camp, the teachers and Natalie were great. We used 3 of the Coles zip up cooler bags for food and snacks and one box of formula and we had the menus and alternatives all marked up.

All the kids were so excited. And I was anxious and nervous but we have to let go at some stage. On night one, the teachers called me and said everything had gone very well and Soph was happy and full. Overall it was a great success and Sophia said she was full at every meal and had a blast. ***So overall Win Win!*** Here are some photos of Soph and Kohen and friends. I have also sent through the Spreadsheet and mark-ups to the MDDA if anyone would like a copy to use for their kids school camps please contact the office.

Paige Moore
MDDA Treasurer

My son Mark - PKU & all grown up!

Mark was born in January 1978 in Central Qld, with PKU, and the only one within 500kms. It was well before mobile phones, internet, and the wonders of the now taken for granted technology. Communications with Doctors & Dietitians were only by post or home phones (and those expensive trunk line calls). Mark attended his first PKU clinic in Brisbane at the age of 8 years, this also included the Xmas party.

As a baby of 5 weeks old (it took this long to send and receive heel prick results), he was started on Lofenalac milk formula and progressed to a combination of Aminogran Food Supplement and Aminogran Mineral mix, Ketovite liquid and PKU tablets. In 1988 he started on Maximum XP, which was great as this was more of a complete formula.

Mark started Grade 1 in 1984, at the small school in the farming area where my husband and I still live. There were under 40 students in the entire school throughout his primary education, which I think made it easier to control his diet and educate his fellow students on the "he can't have" items. *We focused on learning, play and sports, not food.*

He went on to attend Rockhampton State High School, with over 1,000 students, and by then he knew what he could and couldn't have. Mark successfully completed Year 12 and his favourite sport throughout school was rowing in the Fitzroy River, a sport he thoroughly enjoyed. He also got his car and motorbike licence during this time.

Some of Marks achievements are: -

- Senior Certificate – 1995
- Graphic Design/Artist –T.A.F.E – 1997
- Diploma of Graphic Art – 2001
- Bachelor of Social Science @ QUT -2006
- Graduate Diploma Psychological Science – CQU 2015
- Masters of Social Science – Griffith University – 2021.

During his studies he worked as a labourer at a limestone quarry, grounds keeper; life saver at a swimming pool, and has had a small dog washing business.

How amazing the research and developments are that have taken place with the PKU diet since Mark was born. There were no products like pastas, rice, crackers, biscuits, bread etc, or indeed any companies producing dietary products. I was simply given two foolscap pages, one with protein values of fruit and the other of vegetables. In 1986 we were given a prescription for bread mix to be collected from the Rockhampton Base Hospital and I thought this was wonderful.

Mark is now 43 years of age and lives in the Greater Brisbane area, married for over 12 years and has two daughters, neither PKU. He has classic PKU and on 10 grams of protein a day. As you can see I became redundant from the PKU diet many, many years ago when Mark moved to Brisbane in his very early twenties

Regards to all, Janice (Mum).

IEM Food Grant

REMINDER: *Patients with* ***Phenylketonuria (PKU) must reapply for the IEM grant every twelve months.*** *An application form signed by your metabolic specialist must be provided to the Department. Contact details and eligibility information can be found at the link below.*

The IEM food grant payments are due by the Friday of the first full week of each month.

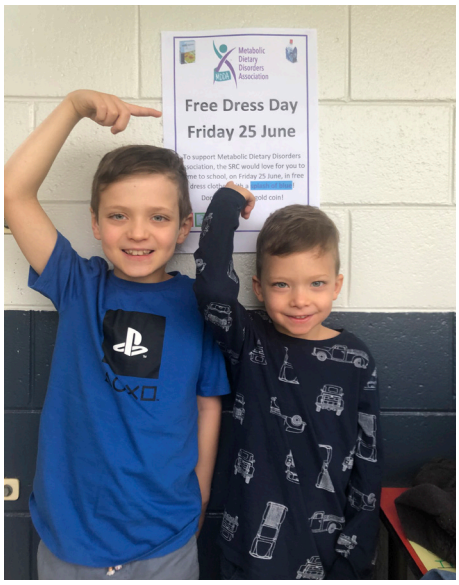
Payment Schedule 2021
10 December

Payment Schedule 2022
7 January, 11 February, 11 March, 8 April, 6 May, 10 June, 8 July, 5 August, 9 September, 7 October, 11 November, 9 December

Visit mdda.org.au/resources/iem-food-grant/for more information

MDDA's young Ambassador, helping to educate and fundraise.

NSW



It is inspiring to hear of how our younger members are taking it upon themselves to raise awareness in their own communities and educate those around them.

A huge thank you to our MDDA member, Sam, his family, and staff and students at Albany Hills State School for their support and fundraising efforts for the MDDA. They raised $1172.00 at the schools free dress day on the 25th June 2021.

This was such a wonderful idea and MDDA are grateful to be the charity that was chosen by Albany Hills State School and appreciative of all the work that Sam and his family put into making this happen. Thank you Sam for bringing this idea to fruition!

Below is the newsletter article that appeared in their School Herald and beside is the speech that Sam and his friend, Olivia, gave in front of the entire school.

It is through fundraising and donations that MDDA can continue towards achieving our vision that all individuals living with an Inborn Error of Metabolism(IEM) are leading a life at full potential, not limited by choice or resource.

SRC News…

FREE DRESS DAY – FRIDAY 25 JUNE – WEAR SOME BLUE

I am allowed 7.5g of protein per day which is the equivalent of a glass of full cream milk. As a comparison, a Big Mac, fries and a thickshake is 56g of protein which is 8 times my daily allowance! I need to measure and calculate everything I eat and use low protein flour, rice, pasta and milk. I also have a special drink that provides the nutrients that my diet doesn't which we call my muscle juice.

Having PKU can be challenging but the support of the MDDA helps me meet other children with PKU through Christmas parties, youth camps, cooking classes and annual retreats so we can have fun and enjoy our low protein food together. The MDDA also raises funds for medical research, educates and supports families living with IEMs.

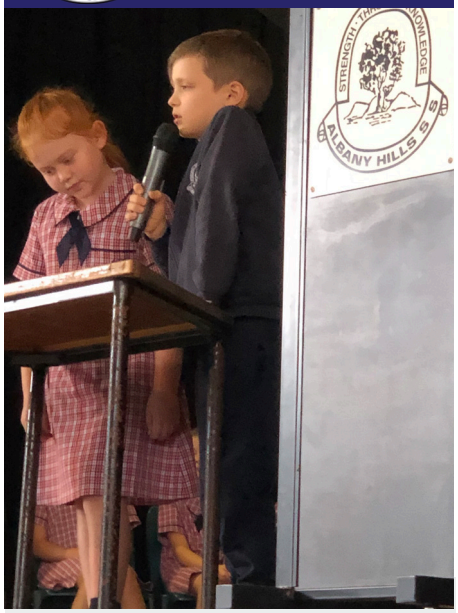
It means a lot to know that my school and friends support me and other kids and adults living with metabolic disorders. Please show your support for the MDDA by wearing blue and making a gold coin donation this Friday 25 June.

Samuel Leahy 3JW


Samuel Speaking on Assembly

Metabolic Dietary Disorders Association

Albany Hills State School
Strength through knowledge



Hi, I'm Sam and this is Olivia and we're from the SRC. (School Representative Council)

Sam: *When I was born, I was diagnosed with an Inborn Error of Metabolism (IEM) called Phenylketonuria or PKU for short. This means that I am required to be on a strict low protein diet for life as my liver cannot break down one amino acid found in all protein called Phenylalanine. It affects 1 in 15,000 babies born in Australia.*

Olivia: *Having PKU must be challenging, but the support of the MDDA helps Sam meet other children with PKU through Christmas parties, youth camps, cooking classes and annual retreats so he can have fun and enjoy his low protein food with others. The MDDA also raises funds for medical research and educates and supports families living with IEMs.*

Sam: *It means a lot to know that my school and friends support me and other kids and adults living with metabolic disorders. Please show your support for the MDDA by wearing some blue and making a gold coin donation this Friday 25th June.*

Thank you ***(together)***

ACT

Thank you to Dominique and her work collegues, who organised and supported her Workplace fundraiser on International Rare Disease day.

#teamMDDA is a way for you and your family and friends to raise funds in support of the MDDA. You can do so by selecting MDDA as your chosen charity as you participate in an activity of your choice and raise awareness and funds for MDDA whilst doing something you love.

Funds raised via #teamMDDA go straight towards providing greater resources and value back into the MDDA community and research priorities.

By signing up to a #teamMDDA challenge you are joining a group of remarkable individuals who are prepared to go above and beyond to achieve something for a great cause. If you are doing a fun run, marathon or other outdoor challenge, #teamMDDA running singlets & T-shirts can be ordered (and personalised) via MDDA Marketplace.

visit www.mdda.org.au/help/teammdda and joining the team

Welcome to one of our newest PKU members – Caleb and his family.

We are always glad to see our MDDA teddies are such a favourite with our newbies.

MDDA have recently setup a new private facebook group for our parents with children diagnosed with an IEM 2years and under. If you would like to join please contact the office for the link. If your child is in this age bracket please let us know if you have not received a PeeKabU Newborn starter kit through your clinic or when you joined (email : office@mdda.org.au) and we shall get one sent out to you!

MDDA would like to thank our major bag sponsor NUTRICIA LIFE-TRANSFORMING NUTRITION

Zest Adult Wellbeing and Vitality Program

Zest aims to improve the mental, emotional and physical health of Australian adults living with an IEM. The program is delivered via MDDA's online virtual events platform via a combination of web-based seminars, teleconferences, online forums, weekly goal setting and one-on-one coaching. The program is facilitated by Louise Healy.

This year we ran an inaugural YOUNG ADULTS ZEST series. It was great to have those that completed the "Introduction Mindfulness for uncertain times" move onto the Advanced group to consolidate their Mindfulness practises.

If you would like to register your interest for a future Zest series or would like more information please email: office@mdda.org.au

MDDA thank our sponsors BioMarin and Vitaflo who support the Zest program. Without their support we would be unable to offer this exceptional program.

Zest

ZONE • ENERGISE • SOAR • THRIVE

BIOMARIN

Vitaflo
Enhancing Lives Together
A Nestlé Health Science Company

Supplier Christmas Trading information

CHRISTMAS HOLIDAY CLOSURE

Vitaflo Australia will be closed from
Thursday 23rd December 2021 and
reopening on **Tuesday 4th January 2022.**
Please ensure your customers have enough
product to cover this period.
For emergency orders please call **03 5229 8222**

Thank you and Merry Christmas!

Vitaflo
Enhancing Lives Together
A Nestlé Health Science Company

Orpharma will be closed from
Wednesday, 22nd December until Tuesday, 4th January.

To avoid any delays or delivery issues over the busy Christmas period, we recommend placing any orders with us for supplements or food products at least 2 weeks in advance.

Orpharma

Cortex Health will be closing for Christmas from ***Wednesday 22nd Dec until Tuesday 4th Jan 2022***

Skeleton staff will be operating infrequently during these times to process urgent orders however all orders will probably take extra time to process and deliver.

We encourage all formula and low protein food orders to be submitted by ***Monday 13th Dec 2021*** to allow sufficient time to get to all during what will undoubtedly be a very busy time.

cortexhealth
Find your balance

NUTRICIA
LIFE-TRANSFORMING NUTRITION

As this is an extremely busy period for customer orders, Nutricia would like to encourage customers to place their orders well in advance to ensure delivery before the Christmas break, and to allow sufficient stock to cover until early in the New Year. *Due to the increased volume of deliveries occurring throughout the national transport system over this time and ongoing delays due to intermittent COVID shutdowns, we highly recommend placing your orders before close of business* ***Wednesday the 8th of December to ensure delivery before the 24th of December.***

Orders placed between the 9th December 2021 and 4th January 2022 will continue to be processed, however may experience extended lead times. Please note, Nutricia's Customer Experience team will have altered opening hours during this period. Below outlines the closure times.
Friday 24th December 2021 - Closed from 12 noon (AEST) Monday 27th -Tuesday 28th December 2021 - Closed
Friday 31st December 2021 – Closed from 12 noon (AEST) Monday 3rd January 2022 - Closed
At all other times our Customer Experience Team will be operating as usual (9am – 5pm AEST).

SHARE YOUR RECIPE
TO GO INTO THE DRAW TO
WIN
A NUTRIBULLET®

lowproteinconnect.com.au

To enter please scan the QR code or visit www.lowproteinconnect.com.au

MDDA Student Placement

Hi everyone, our names are Sunni, Anishka and Nicolle, and we are Master of Genetic Counselling students at the University of Melbourne. It was a pleasure to work with the MDDA members and IEM community.

We had the opportunity to meet a few of you at Tuesday night Community Hub sessions and we were humbled to be able to hear your stories. We especially enjoyed working on the 'Quality of Life' survey and are so grateful for the in-depth responses we received. Being able to hear your first-hand experiences of living or caring for an individual with an IEM provided us with a new perspective on the impacts of IEMs and the importance and value that support and advocacy groups bring to communities.

We would like to say a special thank you to Jo, Jenny, and Lou, who have been a part of our journey in learning about IEMs and the holistic community MDDA has created. We were touched by their empathy, warmth, and compassion upon sharing their knowledge, resources, and stories, and we greatly appreciate their guidance. This experience will be invaluable to our future career in genetic counselling, where we will have an in-depth understanding of clients' journeys.

THE UNIVERSITY OF MELBOURNE

*Calculate all recipes as per guided by your dietitian

Sweet Potato & Citrus Salad

Ingredients:

1kg orange sweet potato, peeled
2 onions cut into thin wedges
2 tablespoons brown sugar
1 teaspoon green peppercorns
¼ cup olive oil
Juice and rind from 1 orange
A few sprigs of dill or parsley

Method:

Preheat oven to 180°C. Line an oven tray with baking paper.

Cut sweet potato and onion into desired wedges and place on the prepared tray. Sprinkle with brown sugar.

Bake for 35 minutes or until tender.

Put sweet potato and onion in a large bowl.

Place green peppercorns, olive oil, orange juice and rind, and dill in a small bowl. Whisk together.

Add the citrus dressing to the sweet potato salad and toss to combine.

Vegetable Chow Mein

MAKES: 1 serving

Ingredients:

1 tablespoon olive oil
½ onion chopped
1 clove garlic crushed
1 carrot peeled & sliced
1 cup cabbage finely shredded
75g uncooked low protein rice
1 cup vegetable stock (1 teaspoon vegetable stock powder + 1 cup water)
½ teaspoon curry powder
Salt and pepper

Method:

Heat oil in a non stick saucepan, and cook carrot, onion and garlic until onion is soft.

Add rice to onion and cook for about 2 minutes.

Add curry powder to stock, and then add stock mixture and cabbage to saucepan with onion and rice, stir to combine.

Season with salt and pepper and place lid on saucepan and simmer for 20 minutes, stirring occasionally.
Remove from heat. Serve immediately and enjoy!

Apricot Muffins

Ingredients:

1 cup water
1 cup sugar
1 tablespoon Nuttelex
2 teaspoons baking powder
250g of low protein baking mix
1 teaspoon of mixed spice
170g dried apricots
1 x mini muffin pan (well greased)

Method:

Bring to boil water, sugar, apricots and Nuttelex for five minutes.

Once boiled add baking powder and stir lightly.

Sift flour and spice and whilst boiled mixture is still fairly hot fold in flour and spice.

Spoon into mini muffin pans and bake in moderate oven (180°C) for 10 -15 minutes or until muffins set to touch and slightly coloured.

Christmas Pasta Salad

Ingredients:

½ box (250grams) low protein pasta spirals, cooked and cooled
1/2 cup sliced sun-dried tomatoes
1 large carrot, julienne sliced, raw
1/3 cup green peas, cooked
2 tablespoons basil pesto
100ml French dressing

Method:

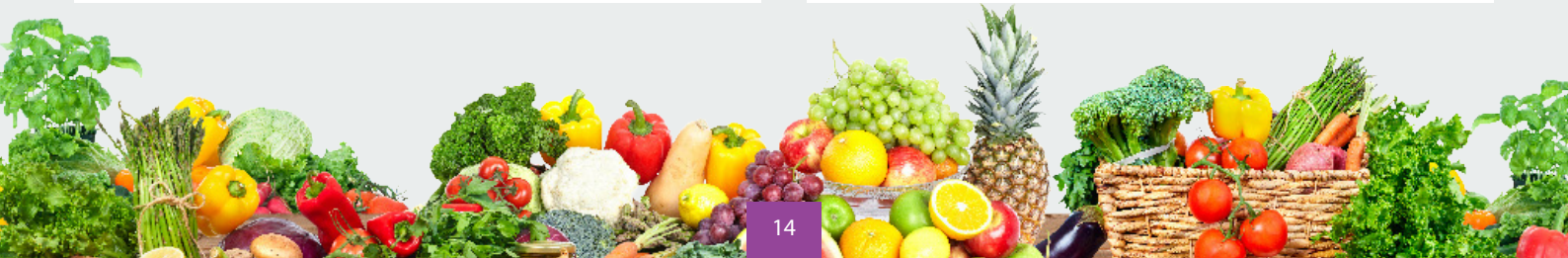
Cook low protein pasta as directed, cool and set aside.

Cook green peas. Cool and set aside.

Mix the French dressing and pesto together in a bowl.

Add the pasta and remaining ingredients to the dressing mixture and mix through.

Transfer to a serving bowl and enjoy!



Products

Always check the nutritional panel on products

Country Life:
Gluten Free & Dairy Free
White Bread
Protein per100g: 2.7g
Protein per serve: 1.8g
Serving size:67g (2 slices)
Available from: Coles & Woolworths

Kez's Kitchen: *Gluten Free*
Lemon Cream Melting Moments
Protein per 100g: 0.6g
Protein per serve: 0.2g
Serving size: 32g
Available from: Coles, Woolworths, IGA stores

Coles: *I'm free from*
Garlic Bread
Protein per 100g: 1.9g
Protein per serve:<1g
Serving size: 50g
Available from: Coles

Natural Pulse :
Italian Sundried Tomato Vegan Sausages
Protein per 100g: 2.8g
Protein per serve: 1.4g
Serving size: 50g
Available online: Harrisfarm.com.au

Genius: *Gluten Free*
Farmhouse Brown Loaf
Protein per 100g: 2.1g
Protein per serve: 1.5g
Serving size: 72g
Available from: Coles

Fruit-tella: *Gelatine Free*
Sour Wrigglers
Protein per 100g: 0g
Available from: Woolworths

Arnott's: *Gluten free*
Tiny Teddies Choc Chip
Protein per 100g: 4.8g
Protein per serve: 1.2g
Serving size: 24g(12 biscuits)
Available from: Woolworths

Liberate: *Gluten free*
Traditional Honeycomb Crumpets
Protein per 100g: 0.9g
Protein per serve: 0.5g
Serving size: 60g
Available from: Coles
Online:Happy Tummies

Woolworths: Banana Blossom in water
Protein per 100g: 1.4g
Protein per serve: 1g
Serving size: 75g
Available from: Woolworths

Special Medical Foods Suppliers

Nutricia/Loprofin
www.pkuconnect.com.au/product-category/low-protein-foods/
Ph: 1800 889 480

Vitaflo / Fate
www.nestlehealthscience.com.au/vitaflo/v2u/low-protein-foods
Ph: (03) 5229 8222

Cortex Health / Cambrooke
www.cortexhealth.com.au/products/low-protein-foods
Ph: 1800 367 758

Orpharma/Mevalia
www.orpharma.com/products/low-protein-food/
Ph: 03 9863 7267

Platypus Foods
www.platypusfoods.com.au

Grab yourself some merch, IEM products or gifts at

MDDA Marketplace

Looking for gift ideas, while helping support the MDDA?
Check out our online shop at MDDA marketplace for some great ideas.
www.mdda.org.au/marketplace

ARE YOU A FINANCIAL MDDA MEMBER?

Please contact us office@mdda.org.au if you would like to check on your status and continue to support the MDDA

MDDA Membership only $35 per year.

Membership ensures MDDA can continue to support all individuals living with an IEM and help them achieve positive health outcomes

- provide more informed choices, resources and support
- provide support by way of website, newsletters, recipe resources, Peer support, conferences, family retreats, social activities and many other programs.
- enables MDDA to provide a voice to government and decision makers on all levels, to promote research into inborn errors of metabolism
- to raise awareness to the greater community and to advocate for all individuals and families living with an IEM.

This newsletter will only grow and get better with your input.

Please share your stories, ideas and tips, birthday celebrations and functions with us via office@mdda.org.au

Contact Us

Address
Suite 4, 6 Thomas Brew Lane
Croydon VIC 3136

PO Box 33
Montrose VIC 3765

Phone 03 9723 0600
Freecall 1800 288 460

office@mdda.org.au
www.mdda.org.au

Executive Officer
Jenny Briant
jenny@mdda.org.au

Administration Assistant
Kerry Tulloch
kerry@mdda.org.au

Communications
Isabella Hendricks
isabella@mdda.org.au

Patient Pathways Telehealth Case Management Nurse
Jo Campbell
Ph: 03 9723 0700
pathwaysnurse@mdda.org.au

Committee of Management

President
Monique Cooper
(PKU Parent VIC)
monique@mdda.org.au

Vice-President
Louise Healy
(PKU Parent QLD)
louise@mdda.org.au

Treasurer
Paige Moore
(PKU Parent QLD)
office@mdda.org.au

Secretary/Public Officer
Bianca Albanese
(PKU Adult NSW)
secretary@mdda.org.au

Health & Wellbeing
Susi Hendricks
(PKU Parent VIC)
office@mdda.org.au

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.*

The MDDA is supported by funding from the Australian Government under the Health Peak and Advisory Bodies Program. MDDA acknowledges and thanks the following industry members and sponsors for their ongoing support.

BIOMARIN MENARINI NUTRICIA LIFE-TRANSFORMING NUTRITION Vitaflo Enhancing Lives Together A Nestlé Health Science Company

Donations over $2.00 are tax deductible