



Metabolic
Dietary
Disorders
Association

ANNUAL REPORT 20 22



The Metabolic Dietary Disorders Association inc. (MDDA) offers support services and resources to educate, connect and enable all families living with a protein IEM in Australia.



Reflections

Time to Make Some Noise

This past year has been another one of significant changes, challenges, and uncertainty for everyone. It has been tough these past few years, but together we have continued to navigate this new reality, providing opportunities to reflect, learn and assess priorities. The IEM community has shown tremendous resilience and adaptability in these challenging times.

I am pleased to report that we have had a very productive and constructive year and together we have several achievements. We continued to embrace the virtual online world to keep engaged with our members and stakeholders. The Member Connect Hub grew, bringing members together online including Spotlight series on members, workshops, panels, connect groups and Happy Hour kept our community connected.

The Great Protein Challenge fundraising campaign returned in May/June 2022. An incredible \$41,000 was raised. To every person and team members who took part in the Challenge and their donors –THANK YOU!

Thank you to the many health professionals who willingly and generously supported our MDDA online events this year. We appreciate your time especially during another year where demands on the health system have been enormous.

To our generous sponsors and partners, without your funding and sponsorship we wouldn't be able to offer the level of service, support and programs to the IEM community. Thank you BioMarin, Menarini, Nutricia, Vitaflo, Cortex Health and PTC Therapies. We look forward to continue working with you in the future.

MDDA have a forward thinking, hardworking and skilled Committee of Management who put in many unpaid hours steering and supporting the organisation and advocating for more resources and better recognition for people with IEMs. I would like to acknowledge and recognise the leadership of Louise Healy (Vice President) throughout the Make Some Noise campaign. Louise's dedication and hard work advocating for fair and equitable treatment for all patients with an IEM over many years is truly admirable. Collectively we continue to use our voices and stories to raise awareness to the wider community and provide a detailed picture of living with an IEM and the challenges that affect people's health, wellbeing, income and quality of life.

We would not have been able to achieve any of our successes without an incredible team of dedicated staff, volunteers and supporters. Your participation is one of our most important sources for future planning and strength to our association. I thank everyone in all areas of the organisation who have worked incredibly hard over the last 12 months to deliver and provide meaningful support to our community - through your hard work, people with IEMs are informed and supported.

A key priority for next year will be developing our long-term strategy, assessing further opportunities and priorities and seek to expand further support and action to achieve our vision that all individuals living with an IEM are leading a life at full potential, not limited by choice or resource.

The IEM community is a unique powerful community driven by strength, dedication and commitment, together I am confident that we can achieve whatever we set out to do – Together, let's Make Some Noise!



Monique Cooper
MDDA, President,
PKU Mum



Realising our Vision



Our Vision

All individuals living with an IEM are leading a life at full potential, not limited by choice or resource.



Our Mission

The MDDA educates, connects and enables individuals and families living with an IEM – ensuring more informed choices and a better quality of life.



Our Goal

Achieving positive health outcomes for individuals living with IEMs, and requiring life-long metabolic dietary management skills, resources and support.

As the National peak support body supporting IEM families in Australia, we strive towards delivering upon our strategic priorities, and this year has been one of incredible outcomes.

Our Focus

Our Achievements

IEM FAMILIES Supporting, Educating, Connecting and Enabling	23 New Members	711 Email Subscribers	17 Online patient forums	
	3300+ Voluntary Hours	2800 Support emails & phone calls	1 6 1 Printed Newsletters eNews	
	1 1 Member Surveys Virtual Conference	13 1 PeeKabU bags delivered		
GOVERNMENT Informing, Influencing and Advocating	2 2 Reviews Advisory responses provided	70 70 Resources & and materials developed and distributed 170 170 Requests for information to the community on health policy matters		
HEALTH PROFESSIONALS Collaborating and Educating	90 Medical practitioners attending MDDA education and training sessions	12 12 Group meetings attended		
GENERAL PUBLIC Informing and Engaging	2.6K Facebook page likes 486 Instagram Followers	4 #teamMDDA fundraising initiatives	8 Active FB groups 1095+ Members	22 New Youtube Subscribers and 1575 views.

The Power of Shared Experiences

Over the past 12 months members have shared their IEM journeys with those in our community through our MDDA Matters newsletters and eNews. These heartfelt, inspiring, stories have helped many realise they are not alone in the day to day challenges they face. We have heard from individuals and families living with MMA, PKU adults, kids & a family's journey of the diagnosis of their PKU baby.



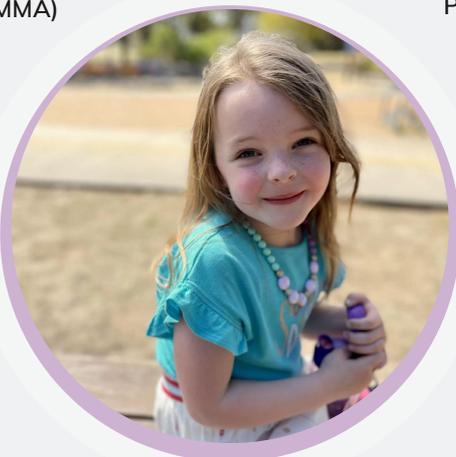
Lachlan
Methylmalonic
acidemia (MMA)
Teen



Sophia
PKU Tween



Mark
PKU Adult



Rosie
PKU child



River
PKU Infant

In Memory of Koby

We are devastated and heartbroken by the tragic loss of our much loved and admired member Koby and two of her beautiful children Chloe and Harrison. Our deepest sympathies extend to her loving partner Craig, and their two sons - Lachlan and Liam. 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 you were one of a kind and will be deeply missed.



Informing government health policy

MDDA assists the Australian Government to inform and support the health agenda by improving linkages, networks and cooperation with our members, the health sector, the wider community and the government. MDDA regularly consults with members and people with lived experience to provide opportunity to inform our policy work and responses to government reviews, strategies and health agenda.

Department of Health

We participate in government consultation opportunities such as Pharmaceutical Benefits Advisory Committee, Life Savings Drug Program, Newborn Screening Working parties, chronic/rare disease forums etc. We coordinate national responses to relevant discussion documents requiring IEM health sector expertise. We provide specific information and knowledge expertise regarding food for special medical purposes. Provide submissions, position statements and other expertise and knowledge to specific government consultation processes to advance IEM care. We identify possible gaps in the Department of Health policy and programs and provide specific advice if detected.

Inborn Errors of Metabolism Program

We work with government to ensure continuity of the IEM Food Grant and keep our members informed of guidelines, procedures, timing of payments, renewal process. In June, MDDA met with new staff in the Infrastructure Programs: Government Partnerships Section and the IEM Program Officer. We provided information to assist in their understanding of inborn errors of metabolism, current treatments, therapies and the role the MDDA play in the IEM Food grant.

Pharmaceutical Benefits Advisory Committee (PBAC)

MDDA provided a submission to the PBAC for the use of Sapropterin for responsive Australian PKU adults and Pegvaliase for PKU patients aged 16yo and over. The basis for our submission is the growing evidence that dietary therapy alone is both unsustainable and does not achieve optimal health outcomes, access to treatments is essential to address these issues. Our submission referenced the importance of equitable access to treatments for all Australians with PKU.

In April MDDA held a PBAC Information Session including Q&A with the PBAC Deputy Chair and consumer Advocate for our members and interested stakeholders. The Deputy Chair explained how the PBAC process works and what information the PBAC finds helpful in the consumer comments they receive for the listing of new treatments.

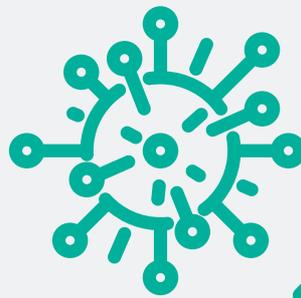
Primary Health and Palliative Care Branch

MDDA participated in an independent evaluation of the Patient Pathways Program. We provided information and advice to assist with the assessment of the pilot program and consider its efficiency, effectiveness and appropriateness.

Newborn Bloodspot Screening program

Newborn bloodspot screening in Australia continues to be known as one of the most successful population health initiatives of our time. This program has meant that our patient population have avoided lifelong disability or death. MDDA promoted the Governments Newborn Bloodspot Screening Program throughout the year.





IEMs and COVID-19

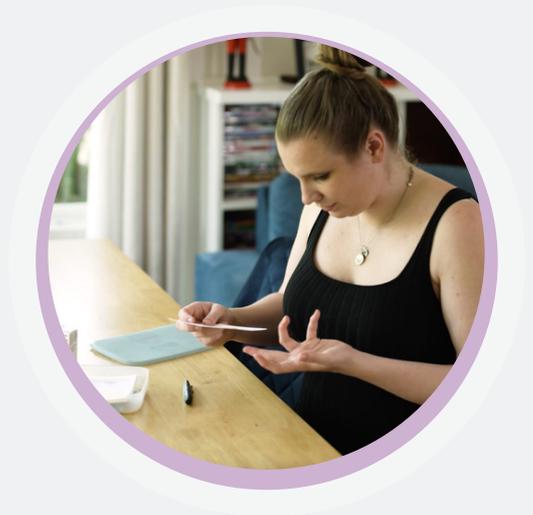
MDDA continued to support the Australian IEM Community providing information and guidance to ensure the needs of IEM patients, those patients at risk and their families and loved ones are supported and informed during the pandemic.

COVID-19 Stories:

We love sharing our members experiences and lived experiences of PKU/IEM families. We shared a series of stories of people and families who have had lived experiences with COVID-19. We hoped by sharing these personal varied experiences, that they may help other families and individuals that may be affected by the virus and raise awareness of the additional challenges that our IEM community face.

MDDA undertook the following work:

- Actively promoted through MDDA communication channels the advice from the Department of Health about vaccination for COVID-19.
- Provided accurate advice on vaccination and IEM healthcare
- Provided guidance and support on mental health impacts of COVID-19 to IEM patients and their families through the Patient Pathways nurse program.
- Provided accurate and up-to-date information regarding the supply of low protein medical foods, supplements and medication



Government Grants

Health Peak & Advisory Bodies Program Grant

MDDA receives funding from the Australian Government to enable MDDA to play our part in informing the Government's health agenda through impartial advice in turn contributing to improved health outcomes for all Australians and their families living with an inborn error of metabolism. This includes consulting and sharing information with our members, the wider health sector, the community and the Government, providing sector knowledge and expertise and providing education and training to health practitioners to improve the quality of health services. The 2022 Federal Budget saw the cancellation of MDDA's funding under the Health Peak and Advisory Bodies Program.

National Disability Conference Initiative (NDCI)

MDDA received funding from the Australian Government under the NDCI. This grant enables MDDA to maximise the inclusion and participation of members and/or their carers to participate in our nationally focused, disability-related, conference.



Patient Pathways Program

The Telehealth Patient Pathways pilot program ended June 2022. This pilot program was funded by the Australian Government co-ordinated by the International Centre for Community – Driven Research. This grant enabled MDDA to extend our services to deliver support to patients and connect them to specialist care, support services and opportunities to participate in clinical trials. MDDA are seeking alternative funding to continue the support the program provided to the IEM community. The service will no longer be available without the additional funding being sourced.



Advancing Care Patient Pathways Program

The Patient Pathways Program (PPP) pilot commenced in September 2018, with patient navigation services provided over three years to 30 June 2022. The pilot aimed to trial a model of care that will help patients of serious and rare diseases connect with the most appropriate health services and treatment.

MDDA were successful grant applicants through a competitive funding application. Over the past 3 years the Program has provided an elevated level of patient outreach support through provision of useful resources and information to patients and families specifically targeted to suit and support their current transitional stage of their IEM journey.



The program provides support to the community in the following areas:

- Assistance with navigating the healthcare system, accessing services and clinical trials.
- Education and information provision.
- Guidance to develop your ability and confidence to proactively manage their condition.
- Social and emotional support for members and/or their families.
- Support of patient care with external healthcare organisations and facilities.
- Support for ongoing patient care with families and carers following hospital admission, discharge, and accident and/or emergency visits.
- Reconnecting adults to clinic and supporting their Return to care and diet.



Patient Pathways Nurse Supporting MDDA Members - Jo

Since the pilot trial has now ended MDDA are currently funding this program on donations and fundraising and have needed to cut the program hours by more than 50%. We are unsure of the sustainability of the program at this time.

At the completion of the PPP MDDA sent out a survey to participants with the following results:

- 96% agreed that the program increased knowledge and access to services, support and treatment pathways
- 90% agreed that the nurse provided beneficial information, resources and support

My family is thrilled that Jo and the MDDA Patient Pathways service was available when our daughter was diagnosed with PKU at birth. We had not heard of PKU before, so had very little idea where to begin with anything. Jo supported us during this time and gave us information to set us up within the PKU community - eg joining online forums and connecting us with other families which was so important for us. Jo has also followed up on our behalf and provided us with information around clinical trials. She is always responsive and efficient. Jo has also been of great assistance in relation to government grants. At a time when we felt very alone, upset and helpless, Jo made our daughter's PKU diagnosis a little more bearable. It is essential that Jo's services are provided to all families in a similar position to ours.

"The patient pathways Telehealth nurse is an essential element of supporting individuals and families with metabolic dietary disorders."

"This service is so important for the IEM community. With such little knowledge of these rare diseases by general practitioners and our reliance being with Metabolic medicine staff who are already overworked. and we have such limited access already to the metabolic team at our hospital."

Number of patients who accessed Pathways nurse

155

Number of Pathways Nurse led referrals/interventions provided

1238

GLOBAL 2021 CONFERENCE

As a founding partner with the Global Association of PKU (GAP), MDDA were the first host nation for the One World, One Vision inaugural Global PKU Patient Conference that took place on Friday 10th - Saturday 11th September 2021. It truly was a great experience that saw people from over 69 countries come together to learn and engage. Taking place virtually this gave the opportunity for people across different continents to hear presentations from experts in their fields.

The event featured live presentations, pre-recorded scientific presentations, documentaries, panel discussions and ways to connect with other PKU people from around the world.

Australia showcased 7 live interactive presentations from experts including:

- Development of a Genetically Modified Probiotic for the Treatment of Phenylketonuria
Prof. John Christodoulou
- Advanced neuroimaging in PKU
Dr. Michel Tchan
- High-throughput screening of potential therapy targets for protein metabolism disorders
Dr. Minal Menezes
- Cognitive, psychiatric and neuroimaging changes in adult PKU patients returning to diet.
Prof. Mark Walterfang
- Emerging research/evidence on co-morbidities and ageing PKU
A/Prof. Gerard De Jong
- Emerging Gene Therapies for Rare Inborn Errors of Protein Metabolism in Australia
Ian Alexander Professor
- Impact of an MDDA Retreat on short-term phenylalanine levels of a sub-group of the Western Australian Adult PKU Population
Catherine Manolikos

Presentations from other world regions included:

- How PKU is Treated in North America
- An update on Gene Therapy in PKU
- PKU Patient Registry
- Why Phe still matters: Changing the view on PKU in the brain
- Nutritional Education Strategies in Chile
- Overcoming the barriers to a better life with PKU: A physician perspective
- Introducing the PKU Diet In An Untreated PKU Patient
- The Forgotten Children of the Model Disease in Peadiatrics
- The Legacy of Prof. Horst Bickel
- Late Diagnosed and Untreated PKU: A Scientific Perspective

PKU RETURN TO CARE

MDDA receive calls from adult PKU patients or family members that have only recently discovered that the current recommendation for PKU is 'Diet for Life' rather than the advice they may have been given in early teens to cease dietary therapy. They are reporting how they, or their relatives, have been impacted by PKU, such as becoming socially isolated, unemployed, having anxiety and depression, and having poor personal relationships.



Keep your Phe in check.

Take control of your PKU and reclaim your seat in life



RETURN TO CARE CAMPAIGN

Sponsored by: **BIOMARIN**
Phenylketonuria | PKU



If you're a PKU patient, balancing your Phe level is a lifelong struggle, and maintaining treatment for life is recommended. If your level goes unchecked and is allowed to slip chances are you are missing out on your best version of life. It doesn't have to be this way. By controlling your Phe level, staying alert to subtle symptoms and actions, you can take back control of your life.

Seek advice from your Doctor or Metabolic Clinic. Contact MDDA to book your **PKU Check** today and get back to your best version of you

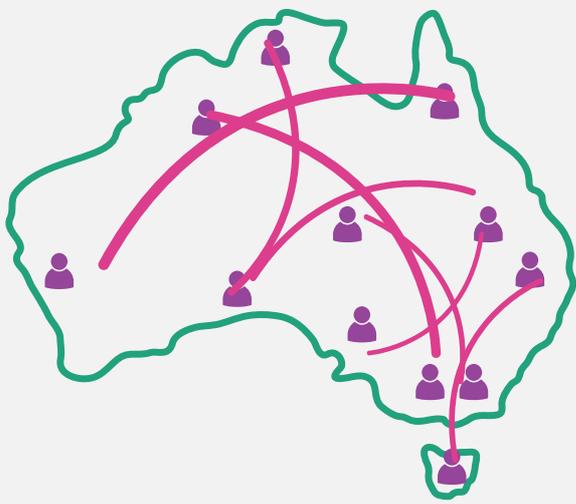
MDDA established a PKU patient outreach campaign to reach as many adult PKU patients, including those that have been lost to the system and those who may be off diet and be unaware of the need to return to diet for a better health outcome as soon as possible. The campaign was designed to raise awareness, advocacy and education of the ongoing burden of disease being faced by PKU adults.

'After 37 years off diet, I made the decision to stay on diet for life. I can honestly say that it is the best decision I have made. PKU does not define me and no longer limits me. Being on diet I can now truly live up to my full potential and enjoy life to the fullest. In life, I have no regrets, however I do regret going off my diet.' Con

'I would like everyone to know the benefits in your health and wellbeing when you return to diet. It was recommended that I resume the PKU diet and formula. After a while I began to feel as if I had more energy and things became a little easier for me.' Susan



Information on the Return to Care compendium and Patient Pathways program <https://mdda.org.au/returntocare/>



MEMBER CONNECT HUB

A welcome place for the IEM community to connect, share and learn

The Hub is an inclusive and interactive virtual platform that is extended to all MDDA members no matter of location as a place to connect and meet with other members, gather in special interest groups and educational forums and attend virtual events and workshops. The Hub provides ability for new and existing members to find people with common interests and conditions and create peer support networks of their own.

The Member Connect Lounge is open 24/7 and provides an interactive, easy to use platform where members can attend educational webinar style presentations, attend group chats, 1on1 panel style interviews with audience interaction and join peer support groups regularly online in an informal setting. The LowPro Inspo Virtual Showcase is an interactive resource where members can find latest information of Low Pro foods, products, recipes, cooking demonstrations and low protein food presentations. MDDA updates take place monthly and provide an interactive forum where members can find out what's been happening at MDDA and ask any questions or raise any concerns.

This platform also provides much needed connections within the community who are feeling isolated and vulnerable due to the pandemic. Thank you to our Hub sponsors, Nutricia, Vitaflo and PTC Therapies for all their support.

Key Member Connect Hub Features

- Member Connect Lounge
- Low Pro Inspo Virtual Showcase
- Educational Connect Forums
- Spotlight Series
- Virtual Happy Hours
- Connect Groups
- Teen Workshops/Panels



<p>A welcome place for the IEM community to connect, share and learn.</p>			
<p>MEMBER CONNECT LOUNGE</p> <p>The Member Connect Lounge is a place where all members can go to discover who's in the MDDA Member and IEM community. Find information and stories about members, writing to share or connect with new and old friends in the community.</p>		<p>LOW PROTEIN SHOWCASE</p> <p>This site provides the latest information on an ever growing resource hub to help you get the most information and use the recipe, products, recipes and much more. Our product partners are actively making our lives easier when the pandemic where they can share and bring us product information, pricing, order times and other resources.</p>	
ADULTS AND PARENTS			
<p>ED CONNECT FORUMS</p> <p>Educational webinar style presentations, followed by Connect Group Chats & Breakouts</p>	<p>SPOTLIGHT SERIES</p> <p>Intimate 1on1 or "on the spot" 1on1 style member interviews with audience interaction</p>	<p>VIRTUAL HAPPY HOUR</p> <p>Casual Virtual Networking through the Member Connect Lounge and various Chat Rooms and our Connect 4 Speed networking</p>	<p>CONNECT GROUPS</p> <p>Special interest peer support groups that connect regularly online in an informal setting</p>
1.5 HOURS + NETWORKING 8:30PM TO 10:00PM TUESDAYS BI-MONTHLY TOPICS INCLUDE: NUTRITION, PTA, ANU CAREERS, SPECIFIC DIETS	45 MINS + NETWORKING 7:30PM TO 8:30PM THURSDAYS (1 PER MONTH) AGENTS INCLUDE: SCHEDULE TO BE ANNOUNCED SHORTLY	1 HOUR 7:30PM TO 8:30PM THURSDAYS (1 PER MONTH) CHAT ROOMS INCLUDE: HOSTED BY MEMBERS AS REQUESTED OR REQUESTED	1 HOUR + NETWORKING 8:30PM TO 10:00PM TUESDAYS (1 PER MONTH) SPECIAL AGENTS INCLUDE: NUTRITION, PTA, ANU CAREERS, SPECIFIC DIETS, AND FINANCE, AND MORE TO BE ANNOUNCED SHORTLY
KIDS AND TEENS			
<p>TEENS LEADERSHIP WORKSHOPS</p> <p>Our Low Pro teens leadership team get together for coaching and guidance to support their IEM management and guide our Peekaboo kids with mentoring skills</p>	<p>TEENS PANELS</p> <p>Designed for Parents and other teens to come and ask questions of various IEM Teens</p>	<p>CONNECT GROUPS</p> <p>Peekaboo Kids Mentoring Groups with Low Pro Teens (monthly challenges & show & tell)</p>	
1 HOUR 7:30PM TO 8:30PM THURSDAYS (1 PER SCHOOL HOLD) TEEN LEADERS INCLUDE: LOW PRO TEENS IEM CAN PARTICIPATE (ONLY PARTICIPATED IN A PANEL) AND ALLOCATED TO A MENTORING CONNECT GROUP	45 MINS + NETWORKING 7:30PM TO 8:30PM TUESDAYS (1 PER SCHOOL HOLD) PANELS INCLUDE: ANY LOW PRO TEENS CAN PARTICIPATE	1 HOUR INC. BUDDY TIME 7:30PM TO 8:30PM TUESDAYS (1 PER MONTH) GROUPS WITH MENTORING LEADERS UP TO A TEENAGE AGE	

Thank you to our Connect Hub Sponsors!



PLATINUM



GOLD



Enhancing Lives Together
A Nestlé Health Science Company

GOLD

Ensuring treatment access for all Australians with PKU

Our 3-month campaign MAKE SOME NOISE

MDDA's position is that all Australians living with PKU should have access to all treatments that have been approved as safe and effective by the Therapeutic Goods Administration (TGA).

**WHY IS IT;
IMPORTANT?
HARD?
UNFAIR?**

MDDA supports access to any new treatment that has been approved to help manage this serious and rare condition. In the lead up to the PBAC's July 2022 Meeting, MDDA led a campaign strongly advocating for both sapropterin dihydrochloride and pegvaliase to be listed on the PBS for adults with PKU.

**RAISE YOUR
VOICE TO THE
PBAC**

Over 65% say PKU has affected their social relationships

Over 90% find PKU restricts their life

Over the past few years, we've seen advancements in the treatment of PKU in Australia, BUT challenges and inequities remain for PKU Adults. Our advocacy campaign highlighted the need that PKU Adults need subsidised access to all treatments that are safe, effective and registered for use in Australia.

**MAKE
SOME
NOISE**

**SIGN UP FOR
THE GREAT
PROTEIN
CHALLENGE**

This campaign is about highlighting the inequities that come with having a rare disease such as PKU and not having access to all available treatments to help ease the burden. Whilst this is not the norm for all people living with PKU, it is certainly a very true representation for many that are struggling with the day to day management and living with their PKU.

**RAISE YOUR
VOICE TO YOUR
LOCAL FEDERAL
MP**

Living with PKU is more than just being careful about what you eat. The effort it takes to stay on diet is invisible, the day-to-day treatment avoids something horrible, brain damage... In Australia, the only currently available treatment option for many PKU Adults is through adhering to a strict, lifelong, low-protein diet.

**RAISE YOUR
VOICE TO YOUR
LOCAL MEDIA**

Over 55% say PKU impacts their ability to be effective in school/work /university

**Just because it's rare,
it doesn't have to be unfair.**



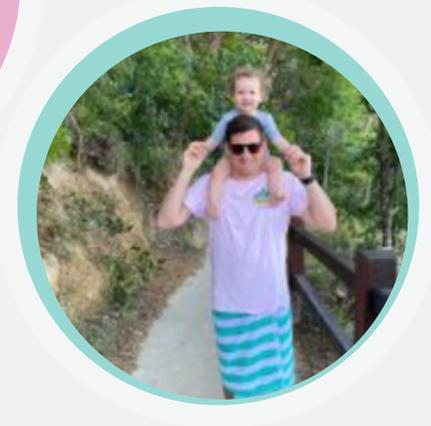
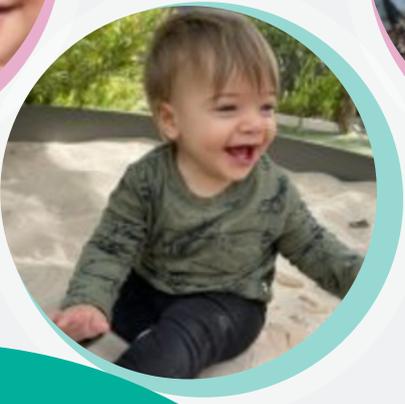
MAY 2022

THE GREAT PROTEIN CHALLENGE AUSTRALIA

Sign up now to PLAY or PLEDGE!
thegreatproteinchallenge.com.au

HOW #LOWPRO CAN YOU GO?

In May and June 2022, over 35 individuals and 9 teams took part in the Great Protein Challenge, walking in the shoes of a PKU/IEM person for a day to understand the importance, complexity and difficulty of managing the disease. The challenge was aimed to eat no more than 10 grams of protein in a day which is MORE than double the amount a classic PKU individual can tolerate in a day.



'On Tuesday 31 May, I participated in The Great Protein Challenge, restricting myself to 6g of protein (Hudson's daily allowance) in support of the amazing work done by the MDDA. Words can't describe how grateful we are for all they have done for us personally this last year, and all they do in the daily fight for fairness for Australians living with rare metabolic conditions.' Yolanda, PKU Mum

The MDDA would like to thank all those who both played and pledged in the 2022 Great Protein Challenge and acknowledge everyone who contributed towards our amazing tally of \$41,500! A special thanks to BioMarin, Nutricia and Vitaflo for their donations. We are excited and looking forward to seeing what we can achieve in 2023!

LowPro inspo

LowPro Inspo is our Low Protein YouTube channel that is designed to provide daily inspiration for people living with protein IEMs.



MDDA Marketplace is the hub for all IEM products, merchandise and gifts. Whether you want to don a PeeKabU hat, or purchase a compendium, all of our many great products are available online to purchase.

#team MDDA

#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.



my LOW PRO pal

To make life a little easier for those managing an IEM, whether it is assisting in protein counting, planning your next meal or helping you to create your next shopping list. MyLowProPal is here to help you and motivate you to stay on track and provide a little bit more organisation and wholistic healthiness into your life.



PTC Member Advantage MDDA

Signing up to the MDDA Member Advantage Program is simple, and costs as little as \$50 per year for a family subscription. What's more, the first year is free for newly diagnosed families – as we understand more than anyone the importance of being a part of a caring and informed support network.

The MDDA Patient Pathways Program is an outreach support system that provides support, mentoring, educational programs and resources for individuals and families living with an IEM throughout every step of their journey.

Zest is the MDDA IEM Adult Wellbeing and Vitality Program. Zest aims to improve the mental, emotional and physical health of Australian adults living with an IEM. Delivered via a combination of web-based seminars, teleconferences, online forums, weekly goal setting and one-on-one coaching.

LowPro PeekabU TEENS kids



Zest



Achieving greater IEM patient health outcomes

MDDA remain in close communication to the nationally recognised Australasian Society for Inborn Errors of Metabolism (ASIAM) to facilitate access to and development of 'world class' best practice management and care for the treatment of IEMs. We regularly communicate with state based metabolic clinics to facilitate discussions for education on improving metabolic healthcare services, strengthening evidence-based policy advice, improving program management, research, regulation and partnerships with other government agencies, consumers and stakeholders.

National Treatment and Clinical Best Practice Guidelines

MDDA remain in discussions with ASIAM for the consideration to develop consistent national treatment guidelines for other inborn errors of protein metabolism similar to those developed for PKU. The MDDA see the guidelines as an important tool for patients to help ask informed questions about treatment and discuss options with clinicians. In addition, we have requested ASIAM also consider developing clinical best practice guidelines. These guidelines would assist with advocacy to help address a lack of clinical resources in certain states in some Australian clinics. We continued discussions with ASIAM on the national guidelines for the testing protocol of BH4, trialling and commencing BH4 therapy for eligible PKU individuals. MDDA are collaborating with ASIAM on an Australia-wide project aiming to collect data from the PKU newborn screened population over 18 years of age to inform health care providers and the Australian government about the current status of the health and needs of the PKU patients in Australia.

Rare Metabolic Disease Workforce White Paper: Towards a Strengthened Rare Disease Workforce for Australia

MDDA attended the webinar launch event of the Rare Metabolic Disease Workforce White Paper: Towards a Strengthened Rare Disease Workforce for Australia. MDDA distributed the Patient and Carer Experience Survey to our members who are represented in the white paper. We also participated as Patient Group Leaders and provided a submission which became part of the qualitative dataset.

The National Strategic Action Plan for Rare Diseases

MDDA continue to collaborate with Rare Voices Australia (RVA), our partner organisation and key stakeholder, on implementation of the National Strategic Action Plan for Rare Disease as it relates to the rare IEM community. In particular on equitable access to treatments, measures to encourage and support clinical trials for rare conditions to occur in Australia, providing input in a rare disease workforce project focussed on metabolic conditions and providing condition specific input in the develop of the multi-stakeholder digital rare disease portal funded by the Australian government.

Member Healthcare Surveys

In July we conducted a PKU Patient Quality of Life survey to better understand the current challenges and future expectations of the patient group. The information collected will contribute to important decisions being made for the PKU Australian community. We plan to use the results of this survey to help inform research priorities, discussions with government decision makers and advocating for access to clinical trials, all available treatments and research funding.

Clinical Trials

We are promoting Australia as a good destination for promising clinical trials. We advocate for compassionate access to treatments for those to participate in clinical trials that are effective with pharmaceutical companies currently in the process of trials. MDDA has worked with other companies considering clinical trials in Australia providing advice regarding regulatory requirements and assistance in establishing clinical relationships. We have provided our members with Australian Government clinical trials information to assist them in decision making regarding participation in clinical trials.

The University of Sydney School of Medicine

MDDA recorded an interview on receiving a Newborn Screening diagnosis for The University of Sydney School of Medicine students. MDDA spoke about how the diagnosis is delivered to parents, the ongoing uncertainty that parents face having a child with a rare disease and their on-going health needs. MDDA highlighted the need for a working partnership between health professionals and patients to achieve optimum health and wellbeing.

Treatment Options

MDDA continue to pursue availability and access to medications and treatments for all IEM patients. Our focus is to ensure that appropriate information is available to families about medications and treatments and to ensure we are in the best position to provide information to policy makers about potential treatment options for metabolic disorders.

Providing patient centred education and resources

Newborn Diagnosis Support

Eleven “PeeKabU” patient support bags have been distributed this year to newly diagnosed families. Coping with the diagnosis of an IEM can be emotional and worrying time, these bags provide a range of management tools and information including experiences from other families that have been through the process e.g. Dealing with and accepting the diagnosis, How do I learn about my babies IEM? What support is there? Infant feeding, blood sampling, family link outreach and mentoring opportunities etc.

Master of Genetic Counselling Students

As part of our commitment to training and supporting health care workers throughout July MDDA began a 3-month host period for three first year Master of Genetic Counselling students from the University of Melbourne for community-based placement. This placement enables the training counsellors to understand the complexities of living with a genetic condition and provided the students with an understanding of the vital role that MDDA provide as a community-based support and service organisation that delivers a continuum of care to people outside of the formal clinical setting and provide many of the psychosocial needs of people. Within this program MDDA presented to the class of first year Master of Genetic Counselling students explaining the difficulties and complex treatment of an IEM diagnosis, providing opportunity for questions.

“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 support and advocacy groups bring to communities. This experience will be invaluable to our future career in genetic counselling, where we will have an in-depth understanding of client’s journeys.” Sunni, Anishka, Nicolle

Zest Adult Wellbeing and Vitality Program

Two Zest programs were run this year in October and November. We held our first program for young adults (18-25yo). This group has been identified as having different concerns and needs to the adult PKU/IEM population. Zest is a purposefully designed and tailored IEM Adult Wellbeing and Vitality program. Research has shown us that more than 70% of adults with an IEM struggle to comply with their complex diet and in addition experience a range of serious co-morbidities including higher rates of anxiety and depression, nutrient deficits, sleep problems, higher rates of obesity and health problems associated with obesity. Zest makes habit-forming life changes in areas identified as limiting individuals from achieving optimal health and management of their IEM. The Zest program provides a holistic and tailored approach to these challenges by addressing emotional, physical and social wellbeing. 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.

Outreach Far North Queensland Metabolic Clinic

MDDA volunteers attended the remote regional metabolic clinics held yearly in Townsville and Cairns. This service has been requested by the QLD metabolic health practitioners who value the support and social inclusion our volunteers bring to the clinic. In conjunction with the clinic the MDDA jointly hosted a dinner for the IEM community to share experiences. This is usually the only time IEM individuals and families living in remote rural areas meet with another person during the year living with the same condition

Pathways Compendiums

MDDA and the Royal Melbourne Hospital are continuing to work together on the Patient Pathways compendium series with the next instalment “Maternal PKU”. This compendium is designed for PKU women planning a pregnancy. Unmanaged maternal PKU can put the unborn child at risk of intellectual disabilities, abnormal brain development, developmental delays, small head size (microcephaly), heart problems and low birth weight. The compendium will include information from preconception through to the birth of baby. We expect this resource to be finalised December 2022.

Social Media Channels

We use a range of social media platforms to provide opportunities for members to have input into MDDA direction, activities and support services. These social media channels also provide tools for keeping members informed of relevant issues and for promoting optimal health outcomes through a focus on treatment compliance and general wellbeing.

Two PKU Pathways Programs - Late Diagnosed & Return to Care

The Late Diagnosed and Return to Care PKU Pathways Programs provide primary care support and education through an integrated stepped care approach. The programs disseminate information compiled by the IEM community to a range of health services and providers to encourage best methods to implement a patient centred approach to educate and support late/undiagnosed PKU patients and those PKU patients who have made the decision to return to their PKU management for best possible health outcomes.

Online Resources

Resources are updated on our website for Care & Education to help with patient empowerment through education and information provision for health services and the wider community. Examples of online resources are: where to find information on medications, information on artificial sweeteners and which are safe for those with an IEM, information on taking blood samples and the COVID-19 advice resource.



July to September

- Member Connect Hub – 6 Member Interviews, 1 Educational forum
- Participation in the RVA Rare Metabolic Disease Workforce Study
- MDDA survey – Australian PKU Patient Quality of Life July
- Updates to COVID-19 online resource
- Kids to Adults Alliance workshop
- Recorded interview on Newborn Screening diagnosis for Uni Sydney School of Medicine
- Contributed to RVA Rare Awareness Rare Education Portal
- Virtual Teens Panel September
- Global PKU Patient Conference host organisation - Sep 10/11



October to December

- AGM - Nov 9
- Parents of IEM Babies Connect Group
- Zest groups
- Hosted 3 Master of Genetic Counselling students from Melbourne Uni for community placement
- Parliamentary Enquiry Report Tabled Nov 21
- MDDA Matters Issue #74 published
- Advocacy Priorities Tabled for next 12-18months
- Members stories published
- Contact Registry for IEMs designed
- Zest Programs @2
- 38 New members July to December
- 437 Patient Pathways nurse-led interventions July to December
- 6 PeeKabU patient support bags distributed July to December
- 14 Connect Group sessions held
- 15 Educational Compendiums distributed



January to March

- Rare Disease Day Feb 28
- Launch Make Some Noise campaign March 30
- Member stories recording x 2



April to June

- PBAC Info Session with PBAC Deputy Chair April 19
- International PKU Day Celebrations June 28
- International Neonatal Screening Day June 28
- Great Protein Challenge May
- Outreach Far North Queensland Clinic May
- 12 New members January to June
- 5 PeeKabU patient support bags distributed January to June
- 3 Connect Hub sessions
- PBAC submission Sapropterin and Pegvaliase
- Meetings with IEM Program heads
- Dept Health independent evaluation of Pathways Program
- 16 New members January to June
- 40 Patient Pathways nurse-led interventions

Year at a glance



Collaborating with purpose

MDDA share ideas and inspiration, seeking to find solutions to specific challenges and shared goals that are innovative, practical and achievable. We collaborate with a broad range of stakeholders and likeminded organisations.

Rare Voices Australia (RVA)



MDDA continue to collaborate with Rare Voices Australia, our partner organisation and key stakeholder, on implementation of the National Strategic Action Plan for Rare Disease as it relates to the rare IEM community. In particular on equitable access to treatments, measures to encourage and support clinical trials for rare conditions to occur in Australia, providing input in a rare disease workforce project focussed on metabolic conditions and providing condition specific input in the develop of the multi-stakeholder digital rare disease portal funded by the Australian government. MDDA participated in the Rare Voices 2021 Virtual National Rare Disease Summit. This event provided the opportunity to engage with key stakeholders and parliamentarians to explore what a person-centred approach would look like for the implementation of the National Strategic Action Plan for Rare Disease in practice.

Global Association for PKU (GAP)



MDDA were the host patient group and organiser for the Global Association of PKU's (GAP) inaugural Global PKU Patient Conference that took place online in September 2021. The first of its kind, this event brought the global PKU patient community together to celebrate 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. 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. Australia was proudly represented with live interactive presentations.

Royal Melbourne Hospital (RMH)



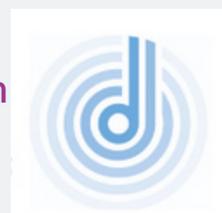
MDDA in collaboration with the Royal Melbourne Hospital launched the PKU Return to Care Compendium resource and program for PKU adults who have difficulty adhering to the diet recommendations due the extent of the dietary protein restriction, lack of access or acceptance of modified low protein foods, poor palatability of medical foods, and the associated costs. It is widely recommended that PKU patients should remain on diet for life for optimal health outcomes. We have begun the 3rd in the Pathways series the PKU Maternal compendium. Planning a pregnancy for women with PKU is essential for the health of both baby and mother.

Genetic Service Network of Victoria (GSNV)



We participate in a range of forums and programs led by the GSNV such as the program Fit for Practice placements for students from the Masters in Genetic Counselling and the Rare Disease Day, Accidental Counselling workshop and other workshops. The support and partnership from the GSNV assists MDDA to increase our capacity to support our members.

Community Driven Research (CCDR)



MDDA are working with CCDR in an integrated approach with the aim to help IEM patients navigate the health system and access all services that are available to them, including clinical trials. The pilot program is in the final year has enabled a specialist tele-health nurse in our organisation to provide a support pathway for patients to access a coordinated comprehensive case management service. The telehealth nurse is available to talk to patients about their chronic health experience and collect a repository of patient experience data that can inform future research, treatment, care, information and support across the health sector. The tele-health nurse role has had an immediate impact assisting in the health literacy and collection of quality data about health and healthcare aimed at managing IEM patients with chronic conditions.

Financials results

30 June 2022

Income 2022

Donations	\$9,546
Fundraising	\$33,946
Grants	\$239,000
Subscriptions	\$1,207
Sponsorship	\$310,527
Other	\$30,122
Total Income	\$624,348

Expenses 2022

Operating Costs	\$83,462
Secretariat	\$220,413
Member Support/Programs/Events	\$127,554
Other	\$6,149
Total Expenses	\$452,372

Although our statements show a profit this can be attributed to delayed conference and project work yet to be invoiced. A complete financial report prepared by our accountants Paul Money Partners is available on request. Thank you to every member who renews their yearly Membership. This is one of the most important contributions you can make to the MDDA. Thank you to every person who had made a donation. 100% of every dollar from a donation is put back into member programs.

Thank you to all those who participate in #teamMDDA member driven fundraising initiatives; Become a #teamMDDA supporter and raise funds for MDDA as your chosen charity through participating in an activity of your choice. Raise awareness and funds for MDDA whilst doing something you love.



Make a donation

Donations made to MDDA go towards a number of different initiatives or research projects.

MDDA is seeking funding to support a number of research initiatives in the area of progressing knowledge and potential new treatments/cures for PKU and other IEMs. Donations over \$2 are fully tax deductible

Volunteer

Join MDDA and become involved with the committee or project team and be a part of making a difference for all individuals with IEMs whilst making new friends.

Host a Fundraising Event

Hosting a fundraising event can be a fun and rewarding experience and provides a good mechanism for raising awareness and money for IEMs in our community.

Looking to sponsor a program, initiative or event? MDDA has a number of programs and events looking for financial support. Examples include:

- IEM Family Assist - retreats & remote clinics
- My Low Pro PAL
- PKU Pathways - outreach support, mentoring
- National PKU & IEM Research & Clinical Care Fund

Acknowledgements

Sponsorship

Thank you to all of our sponsors for supporting and sharing our vision that all individuals living with an IEM are leading a life at full potential, not limited by choice. We appreciate the support and generosity of our partners and sponsors, without them we would not be able to deliver the current level of programs and resources we offer. Our National Family Retreats and other projects would not be possible without their generous support and contributions. Thank you for your continued collaboration and commitment in supporting the IEM community.



Supporters



MDDA Executive Committee and Team

President

Monique Cooper, PKU Parent VIC

Vice-President

Louise Healy,
PKU Parent QLD

Treasurer

Paige Moore,
PKU Parent FNQLD

Secretary

Bianca Albanese, PKU
Adult NSW

Health & Wellbeing

Susi Hendricks,
PKU Parent VIC

Executive Officer

Jenny Briant

Administration Assistant

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