

# 2020



## Annual Report



Metabolic  
Dietary  
Disorders  
Association

# Reflections

As I take the opportunity to reflect on these past 12 months, I take myself back to our National Family Retreat theme “Time to Shine”, shining a light on everything good that is happening with IEMs today.

As a rare disease patient population, the time has never been greater for us than right now. With many advances in clinical trials, research and genetic science leading the way, presenting new opportunities and the hope for a brighter future for all individuals living with an IEM.

The advocacy work MDDA has been driving with key stakeholders and the Australian government is to ensure protein IEMs and all rare diseases are high on the national health agenda. Our efforts are continuing to open doors for our plight for access to best available treatments and equitable healthcare for all.

March 2020 brought a new meaning to “Time to Shine”, as the MDDA moved swiftly into action when COVID-19 changed the plans for all of us. Our primary concern was to ensure the welfare, support and wellbeing of the IEM community was at the forefront of each decision and action that we took. All whilst ensuring we were assisting with the increased anxiety, member concerns and producing resources for our members and IEM families in relation to the changes impacted by the global pandemic. We liaised with the metabolic clinics as they moved to a full telehealth service. We kept in close contact with our specialised medical low protein food and medicine suppliers. I want to congratulate each of the companies on how well the crisis has been managed with every one of our suppliers working around the clock and at sometimes great expense to ensure the service and needs of our patients were maintained.

The introduction of our Patient Pathways Program could not have come at a better time than in 2020. The 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 own journey.

The program includes different initiatives and services that support those with an IEM through every stage of life, from first diagnosis, through childhood to adulthood. This Pathways program aims to develop skills, knowledge, motivation and build on the network of peers for all ages to live their best life possible. Within the program we facilitated our Zest Wellbeing Program, Teen Transitional Workshop and the Patient Pathways tele-health nurse program. You'll find further details on each of these programs in the following pages.

Thank you to every volunteer in all areas of the organisation who have worked incredibly hard over this challenging year. Your participation and support is one of our most important sources for future planning and strength to our association. I look forward to another year building on the work we've completed and continuing to work with, and for, the community of people in Australia living with an IEM. 2021 will see MDDA collectively 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.

Whilst for many of us the challenges we face daily are still very real, I have found time to embrace and celebrate everything that shines and together let's inspire and lift each other into a brand-new shiny year!



A stylized, handwritten signature in black ink.

**Monique Cooper**  
MDDA, President

# 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

<b>IEM FAMILIES</b>  Supporting, Educating, Connecting and Enabling	 <b>27</b> New Members	 <b>788</b> Subscribers	 <b>980</b> Online patient forum participants
	 <b>3700+</b> Voluntary Hours	 <b>3800+</b> support emails & phone calls	 <b>1</b> Printed Newsletters
	 <b>5</b> Online eNews	 1 National retreat – over <b>140</b> attendees	 <b>16</b> PeeKabU bags delivered
<b>GOVERNMENT</b>  Informing, Influencing and Advocating	 <b>10</b> Reviews	 <b>70</b> Resources & and materials developed and distributed	
	 <b>10</b> Advisory responses provided	 <b>32</b> Requests for information to the community on health policy matters	
<b>HEALTH PROFESSIONALS</b>  Collaborating and Educating	 <b>135+</b> Medical practitioners attending MDDA education and training sessions		 <b>53</b> Group meetings attended
<b>GENERAL PUBLIC</b>  Informing and Engaging	 <b>2602</b> MDDA Facebook followers	 <b>9</b> YouTube Videos	 <b>4</b> Advocacy Awareness Videos
			 <b>8</b> #teamMDDA fundraising initiatives

# Empowering our community

## Carmen's journey supported by the MDDA IEM Adult Wellbeing and Vitality Program Zest

I grew up on the Sunshine Coast and lived a relaxed lifestyle with the beach within a short walking distance from my home which served as my playground and my backyard of imagination and curiosity and fun.

I remained on diet until I was a teenager when the advice of the metabolic clinic at the time was if academics were good, social relationships were strong and emerging then it was ok to come off diet, so I did, but little did I know that this was the time in my life (knowing now what we know about the benefits and recommendations today that diet is for life) where I needed diet the most.

Research tells us that more than 70% of adults with an IEM struggle to comply with their diet, in addition they can experience a range of serious co-morbidities including higher rates of anxiety and depression, nutrient deficits, sleep problems, higher rates of obesity and other health problems.

This would be just the beginning of going through challenging times, because life comes in seasons, some are wonderful, and some are hard to move through. This season of the changing teenage years I look back and know that it was the beginning of my journey of learning who I am and who I want to be and where I fit in the world but with the added challenge of being off diet and the deep immense pain of losing a parent made it not such a great season for me. It is safe to say this was one of the hardest and lowest times in my life. But what I know today is that all the experiences and the seasons I lived through has really helped shaped who I am today.

The MDDA IEM Adult Wellbeing and Vitality Program – Zest was developed to support and achieve greater holistic health outcomes for IEM adults, aiming to improve the mental, emotional and physical health of Australian adults living with an IEM.

I remember reaching out to MDDA for support. During this time, I contacted Jo, the patient pathways support and she told me about the Zest Program that was beginning. When she told me about the program,

I remember her explaining it to me as a wellbeing program with a focus on mindfulness during uncertain times. I had not long ago begun my own journey on understanding mindfulness as my psychologist had mentioned it to me and I had become intrigued already but hadn't had any direct coaching or support on how to put it into practice and was struggling to get it right and make it a part of my life, though wanted to know how. I committed to the Zest program completely, 100% all in as I knew that the best benefits would be felt with me being dedicated.

Empowerment  
is the process of  
becoming stronger and  
more confident, gaining  
control of one's life and  
claiming one's rights

(Oxford Dictionary).



The Zest program provides a tailored holistic approach to these challenges by addressing psychological, physical and social wellbeing. The program includes an 8-week coaching forum focusing on healthy goals and habits, learning various tools and strategies to take on challenges and develop a support system and new habits to help you to thrive.

What I really liked about the program was that it was a holistic approach to moving through challenging times. It was not necessarily based on PKU and

the management of this in a mindful way but it was flexible enough that if that was where your struggle lied and were focused on improving at present, it certainly could have been. It was an open program that allowed you to bring your whole self along for the journey. In my situation I had other things I wanted to tackle and knew that the mindfulness tools would set me on my way for holistic healing that would then also enable better management of PKU as result of the benefits from the program.

The Zest program uses modules to get you into the right zone for optimal physical, emotional and social health includes individual goal setting, strategies for developing and maintaining healthy habits and cultivating the peer support network. Using a combination of individual coaching, group sessions and expert presentations. Zest helps identify strengths, passions and strategies to embed for the participants ongoing success.

I absolutely loved the one-on-one coaching. I had looked into this for myself previously and was not able to afford the time of a one-on-one coach. I went really deep with my goals and feelings and self-reflection and dedicated my time and energy to the goals and work in practice that Louise set out for me and lucky for me I reaped the benefits and was able to ultimately in the end make mindfulness an embedded practice within my day to day living. From this I was able to reduce the effects that stress and anxiety was having on my body and through time was able to overcome hurdles using mindfulness as a part of my living that in the past would have sent me into deep anxiety and panic.

Working through the practices of the program allowed me to begin to feel lighter, calmer in my general existence every day and was able to have better responses not reactions to situations. I began to learn to act from a place of self-compassion (I am not perfect at this yet but getting better with practice) and learnt to motivate myself to have success of small achievements by acting from a place of purpose and intention. I was able to commit to embedding meditation and yoga into my daily routines and mindfulness became a practice that was embedded into my days in almost every moment that I was able to. Now don't get me wrong, I am certainly not the mindfulness warrior and boy gees it doesn't always come easy and I have had setbacks where it has not been able to be a part of my days or at least I have not been in the mindset to allow it. But when this happens, in time; I bring myself back to my breath to allow myself to enter into a space of clarity in order to be able remind myself all over again the benefits mindfulness has provided me and how the practice can and will support me now and into the future.

The Zest program and the members from the MDDA committee who developed this program and volunteered their time, effort, care, concern and coaching has gifted me the learning of a practice that I will always now turn to support me in living with an anxious mind and allow it to flourish into a heartfelt mind. This I always be grateful for.

Without the generous financial support from MDDA's valued sponsor the Zest program would not be available – our deepest gratitude and thanks to Vitaflo the founding program sponsor.

## IEMs and COVID-19

COVID-19 saw an increase in the volume of enquiries and support needed through our organisation from the IEM community. As the worldwide pandemic continued the level of anxiety within the community escalated. Our workload increased with members that are case managed which placed an additional load on our systems in relation to the length of time per case as the complexity increased. MDDA will continue to collect COVID-19 data information from our members to be used for future research and studies.

For some rarer IEMs a sick day action plan is commonplace. We distributed information about the precautions to take for individuals with a 'sick day action plan' with regards to COVID-19. MDDA received many questions regarding how this virus may affect those living with an Inborn Error of Metabolism, particularly with issues in obtaining specialised medical food and medical supplies. In order to help with concerns MDDA created a resource on our website to assist with these common questions and provided information and resources to help navigate these challenging times. We encouraged people to keep updated and aware of current developments and notifications from the Federal Government and local State Government.

MDDA provided advice to members regarding Australian Government policy on the distribution of prescription medication during the pandemic. The advice was provided for clarity, reassurance and to ensure that patients could seek medical advice regarding management of their IEM condition.



### Reassurance in uncertain times

Covid-19 - Steps we are taking to help our community feel safe and confident

- Liaise with suppliers to ensure continuity of medicines, supplements and low protein food
- Installing calm to prevent stockpiling of specialised low protein foods
- Created an online resource, providing accurate and relevant information and resources
- Liaise with metabolic clinics and patients as they transfer to tele-health services
- Patient Pathways nurse providing additional support or assistance where needed
- Keeping our community connected
- Continue our mission to educate, connect, inform and support the community throughout the crisis.



## 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

MDDA met with the Department of Health in October to advise the department of our knowledge and experiences of individuals who had recently started on Sapropterin treatment. The first pharmacological treatment for use in Australia for those with PKU. The medicine was listed on PBS in May 2019 for children and adolescents who will continue access when adults under 'grandfathering' arrangements. MDDA continue to access for new therapies and treatments for all ages and all IEMS.

### Life Savings Drug Program (LSDP) Review

As part of the Australian government review of the LSDP for specialised treatments for ultra-rare diseases, each treatment on the program are reviewed every two years. The review offers an opportunity for patients and their families to have input into how the treatment is administered and to provide a patient perspective on the impact of the treatment. MDDA has provided input in the terms of reference for the review and participated in a stakeholder consultation to ensure that the terms of references covered all issues of concern. MDDA distributed information to families for patients to provide the opportunity to respond to the review for specialised treatments for the ultra-rare disease Hereditary Tyrosinaemia Type 1 (HT-1).

## Government Grants

MDDA is funded under the Health Peak and Advisory Bodies Programme.

### Health Peak & Advisory Bodies Programme 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.

### 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 held in Australia.

### Patient Pathways Program

MDDA were successful in receiving funding for the new pilot program funded by the Australian Government provided by the International Centre for Community – Driven Research. This grant enabled MDDA to extend our resources to include a tele-health nurse support service. This service connects patients with specialist care, support services and opportunities to participate in clinical trials.



# Advancing Care Patient Pathways Program

MDDA's Patient Pathways Program provides 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 Pathways Program provides support, mentoring, educational programs and resources for individuals and families throughout every life stage. Within this program MDDA provides a specialist tele-health nurse which provides patients access to a coordinated comprehensive case management service no matter where they live throughout Australia.

The Program provides a pathway for support for patients to access a coordinated comprehensive case management service, to talk about their experience and collect a repository of patient experience data that can inform future research, treatment, care, information and support across the health sector.

This program does not replace the expert clinical care of the patient's metabolic team and does not provide treatment advice. The main objective is to develop tailored care plans, provide practical and emotional support, education, and connection with existing services.

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 your 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 Diet for Life.

The Patient Pathways Program is federally funded by the Australian Department of Health. MDDA are working with the Centre for Community-Driven Research (CCDR) to implement this program. The program aims to increase the capacity of patient organisations to help patients navigate the health system and access all services that are available to them.



"Supporting the IEM community to live their best lives and reach their full potential brings me so much satisfaction. Being able to support and educate our members to become optimal self-managers is one of the reasons I have been nursing for the past 35 years".



Jo

"Working with the IEM community has highlighted for me the need for the wider community to understand the chronic burden of living with an IEM and why it is important for people who live with IEM's that collectively we must influence future research and management of all IEM's in Australia."

"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. I've been inspired by the MDDA Team, they are compassionate change agents nationally and globally. I'm looking forward to 2021 to continue assisting the inspiring and amazing IEM community."



## MDDA Signature events



### Family Retreat

4 - 6 October 2019, Tamar Valley Tasmania

The Annual National Family Retreat saw over 140 members, health professionals, guest speakers and key stakeholders join in the experience. It was the perfect setting for a weekend of learning, connecting and sharing stories with others. "Time to Shine" was the theme at this year's retreat, encouraging us to embrace and celebrate everything that makes us shine and together find our inner sparkle. MDDA wanted to shine a light on everything good that is happening with IEMs today. As a rare disease patient population, the time has never been greater for us than right now.

*"This was my first Retreat, and I didn't know what to expect. After the first few hours I felt comfortable and accepted within the group. I have met some amazing people and some lifelong friends. I have gained so much knowledge that I will now have to contemplate and implement into my life. I hope to attend next year with my husband."*

We are most grateful to have first-class health professionals and researchers who generously give their time to present at our retreats. Their presentations and educational sessions provide invaluable learning experiences.

*"As new parents on the IEM journey the 2019 retreat allowed us to see the amazing future she has and the support that is available for both us and her. Thank you, as new parents this was an emotional roller-coaster and everyone was very supportive and welcoming."*

Our informative sessions this year included the Teen Transitional Workshop providing information describing key components of transitional care. A facilitated session focusing on supporting grandparents who provide extended family support. The 'Zest Boost Camp' included various presentations focused specifically for IEM adults and their partners delivering inspiration and motivation. This session introduced the new Zest program, a comprehensive and personalised program that covers mental, emotional, social and physical wellbeing.

*"I highly recommend retreats to all family members – including grandparents and siblings, great information and inspiration to make living with IEM easier to handle, so much fun and support to last the year through."*







Our PeeKabU Kids Club Program ensured the kids had a weekend full of exciting activities. Not too much encouragement is needed for kids to meet their peers, join in activities and form lifelong friendships.

*“Attending the retreat was life changing for our family. We met families living with the same challenges as us. And our daughter had the opportunity to celebrate having PKU. We left feeling empowered, hopeful, and excited for the future. We left having made life-long friendships.”*



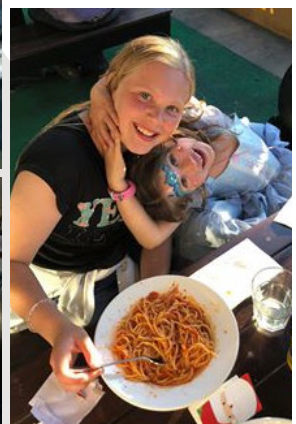
The MDDA thank all the volunteers, presenters, health professionals, sponsors and in-kind partners that all played a key role over the weekend.

*“The scope of what was covered over the weekend was beyond our expectations as newcomers to the IEM community. Meeting other families and such a dedicated group of volunteers and genuine caring people has been a truly life changing moment. We now feel we have so much more love and support as we continue on this journey together.”*

## End of Year Celebrations

Our end of year gatherings provide an opportunity to celebrate the year and reconnect with fellow MDDA members and friends. These events are a great occasion for new families to come along and connect in with the MDDA local community.

State events took place throughout November and December at; Riverside/Tasmania, Ashgrove/Queensland, Wollongong/New South Wales, Calista/Western Australia, Warrandyte/Victoria and Oakden/South Australia





**LowPro Inspo** is our Low Protein YouTube channel that is designed to provide daily inspiration for people living with protein IEM's. The channel initially focuses on low protein cooking and recipes but will expand to also include other inspirational content from MDDA, our members and affiliated communities.

Whether you're a cooking fanatic, or a novice, there is something here for everyone. From cookies to noodle salad to bread, learn new recipes and new tips to cook something deliciously low pro!

LowPro Inspo is open to everyone, why not join up to our Member Advantage program and also become an inspirator, contributing to our inspiring and growing library of content.



All proceeds from the sale of items on the store go towards supporting further MDDA member programs and initiatives.

MDDA Advantage members also gain access to special member offers and discounts.

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



**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 – with all proceeds going to the MDDA to help us continue to support our Member Advantage Programs.

## MDDA CONNECT

**MDDA Connect** delivers communication and support services across multiple platforms and channels. When you first join our community, MDDA Connect is the way that we connect you in with other members so you can learn new information, stay updated and make lifelong friends. Whether you are a subscriber or a part of our Member Advantage Program, you can benefit from the various features and benefits of Member Connect.

MDDA Connect features include:

- MDDA Community and Member Events
- Patient Community Facebook Forum
- Website & Social Channels
- MDDAs 1800 Number
- MDDA Newsletters

## THE GREAT PROTEIN CHALLENGE AUSTRALIA

The Great Protein Challenge was postponed in 2020 due to the unprecedented circumstances of Bushfires and the COVID-19 pandemic. However, we are looking forward to The Great Protein Challenge 2021.

The Great Protein Challenge (now a global awareness campaign) is a public health educational awareness and fundraising initiative that helps support kids and adults living with IEMs. It enables people to walk in the shoes of a person living with an IEM for a day to understand the importance, complexity and difficulty of managing their condition.

By being a part of The Great Protein Challenge, you will help raise awareness for all protein IEMs and provide an informative and fun way to engage in the conversation.

Funds raised through the challenge will go straight into continuing to support the community through building further programs and contributing to MDDA's Research foundation.

HOW #LOWPRO CAN YOU GO?



Thank you to our sponsor

**BIOMARIN**

10

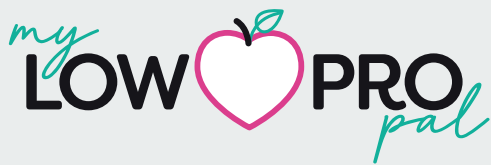


**#teamMDDA** is a way for you and your family and friends to raise funds in support of the MDDA. You can do so by selecting MDDA as your chosen charity as you participate in an activity of your choice and raise awareness and funds for MDDA whilst doing something you love.

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 doing a fun run, marathon or other outdoor challenge, #teamMDDA running singlets & Tshirts can be ordered (and personalised) via MDDA Marketplace.



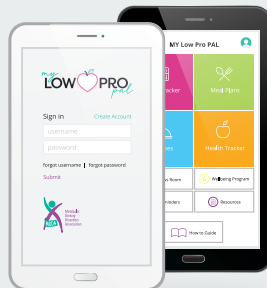


Unfortunately due to the COVID-19 pandemic the development of MyLowProPal has been delayed, but steps are being made to progress the development aiming to release next financial year.

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.

- Protein counting (per Australian IEM guidelines)
- Low protein recipes & meal plans (for all ages & protein allowances)
- Low protein product catalogue
- Shopping lists
- Daily food tracker
- Wellbeing goal setting & monitoring
- Blood spot monitoring
- Reminders

... Plus much more



ZONE • ENERGISE • SOAR • THRIVE

**Thank you to our founding program sponsor VitaFlo**

**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. Zest makes habit-forming life changes in areas identified as limiting individuals from achieving optimal health and management of their IEM.



**Innovation in Nutrition**  
Australia

The initial program includes a coaching forum with 4 modules ...

Zone	Energise	Soar	Thrive
Get into the Zone by focussing on healthy goals and habits.	Be Energised and ready to tackle challenges life throws at you.	Learn various tools and strategies that will help you to Soar.	Gain a support system and new habits to help you Thrive.



**Member Advantage**

By joining MDDA's **Member Advantage Program**, you open the door to another level of support, with access to many specifically designed programs, products and services created to help support families and individuals navigate the pathways of managing their IEM – throughout all of the life stages.

Signing up to the MDDA Member Advantage Program is simple, and costs as little as \$35 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.



**Low Pro Eats** is the MDDA foodies program. Designed to provide greater access and choices to people who Shop and Dine Low Pro.

Featuring:

**LowPro Eats - Shoppers Guide**

A guide that lists online and grocery outlets selling specialised low protein products and other suitable specialty items.

**LowPro Eats -Venue Program**

Enjoy hassle free and inclusive low protein dining out experiences by dining at a recognised Low Pro Eats establishment.

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. The Pathways Program includes different initiatives and programs that support those with an IEM through every stage of life - from first diagnosis to childhood to adulthood.



Parents of newly diagnosed babies, Food starters, Preschooler, School starters, Tweens, Transitioning adolescents, Adults, Maternal women and Carers of high needs (late diagnosed) PKU patients.

Within this program, MDDA provides a specialist tele-health nurse which provides patients with access to a coordinated comprehensive case management service, to talk about their experiences and collect a repository of patient experience data that can inform future research, treatment, care, information and support across the health sector.





## Achieving greater IEM patient health outcomes

MDDA remain in close communication to the nationally recognised Australasian Society for Inborn Errors of Metabolism (ASIEM) to facilitate access to and development of 'world class' best practice management and care for the treatment of IEMs.

### National Treatment and Clinical Best Practice Guidelines

MDDA continued discussions with ASIEM for the consideration to develop consistent national treatment guidelines for other inborn errors of protein metabolism similar to those developed for PKU. In addition, we have requested ASIEM also consider developing clinical best practice guidelines. These guidelines would assist with advocacy to help address a lack of clinical resources in some Australian clinics.

### Research project

MDDA made a formal proposal to ASIEM to instigate a research project to understand the IEM communities' expectations and concerns in relation to emerging treatments for IEMs. The collaborative project will collect survey and focus group data to better understand the unmet need for IEM treatment in our patient community. Data collected will be used in the provision of advice to government in relation to access to medicines and clinical trials.

### Treatment Options

MDDA continue to pursue availability and access to medications and treatments for all IEM patients. As a result, ongoing discussions with ASIEM remain to explore financial support options for research regarding the use of medications for metabolic disorders in Australia. 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.

### Methylmalonic Aciduria (MMA) & Propionic Acidemia (PA) Clinical Trials

The MDDA have been actively seeking information through ASIEM about potential MMA & PA clinical trials in Australia. MDDA distributed information to the overseas phase 2 clinical study for patients with MMA and PA. Currently there are no targeted pharmacologic treatments for MMA or PA that can improve quality of life or extend lifespan. MDDA supported members and provided information to assist with questions on the participation of the trial.



# Providing patient centred education and resources

## Resources

MDDA developed a pilot Transitional Workshop for Young People and Parents/Guardians which includes a downloadable resource to describe and assist in the key component of transitional care for children transitioning from paediatric to adult services. We have identified that this time is where there is high risk of patients being lost to treatment and thus incurring significant preventable health issues. The program aim is to offer practical guidance to help young people and their parents/guardians to understand and prepare for the transitional process. To promote skills in communication, decision making, assertiveness and self-care, to promote independence and provide support and guidance as the young person transitions from their families/carers care to the adult health care service.



Sixteen “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.

MDDA created an on-line downloadable brochure “Clinical Research and Clinical Trials – Information Resource”. This resource is written for the patient to understand the benefits and implications of participating in research and clinical trials, what to think about when considering being part of clinical trials, where to find further resources etc. This resource is soon to be available on our website.

Under development is a Hospital Good Health Tool Kit. This tool kit will be accessed via our website for the hospital treating team to access when a PKU/ IEM patient is admitted to hospital by way of planned or emergency admission. The tool kit resource will promote the importance of adherence to dietary therapy to optimise recovery, reduce days spent in hospital and provide holistic treatment advice.

MDDA developed an online resource for ‘School Starters – Preparing for childcare, preschool and school’, including downloadable resources to describe and assist in the key components of children transitioning to childcare and school. The resource is aimed to support families and offer practical guidance to ensure that the transition for children with IEMs and their parents can be a positive experience and create a positive learning environment. The resource contains information that can be downloaded to assist in planning and creating discussions to ensure the child’s IEM management is understood by their educators.

## Master of Genetic Counselling Students

Two students from the University of Melbourne completed their community placement with the MDDA. The program has been developed to facilitate an understanding of the role of community-based support and service organisations and other health services in the provision of genetic health care. Community placement provides students with an opportunity to consider the broader context of how health and social issues faced by individuals and families with a history of a genetic condition can impact on a person.

*“It was a pleasure for us to witness your sense of community and really confirmed for us the importance of a group, like MDDA, in providing lifelong support to those managing a chronic genetic condition. We want to sincerely thank the MDDA and all MDDA members for welcoming us into your community and allowing us the opportunity to learn from your experiences. Your stories are something we will certainly carry with us throughout our lives and our future practice.” Erin and Anastasia*

# 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)

In October 2019 MDDA joined RVA at a Parliamentary event in Canberra at the launch of the report “Disability & Rare Disease: Towards Person Centred Care for Australians with Rare Diseases”. MDDA contributed to the report which has helped to inform the development of the National Strategic Action Plan.

MDDA attended the RVA launch of the National Strategic Action Plan for Rare Diseases in February 2020. This was a milestone event held at Parliament House, Canberra, with the Minister for Health. The Action Plan provides guidance and direction around key goals and priorities for Australians living with a rare disease. It is the first nationally coordinated effort to address rare disease in Australia and drives systemic reform for universal access to best treatments and health outcomes for people living with rare disease. It is based on three key principles: person-centred, equity of access and sustainable systems and workforce.

MDDA participated in a round table, convened by Rare Voices Australia, to provide input to the LSDP of issues of concern for patients during the COVID-19 pandemic. We sought comment from the community and provided patient comments as feedback to the LSDP secretariat. The secretariat was able to provide comment to us on several matters of concern to patients and this feedback was then relayed to the community.



## Royal Melbourne Hospital (RMH)

MDDA in collaboration with RMH completed the PKU Carer Compendium resource and program for missed or late diagnosed PKU adults living in shared community housing facilities or nursing homes. Restrictions placed upon the community due to the pandemic opened up an opportunity for us to launch the compendium virtually and nationally without limitation by location. The high-quality visual handbooks and personal compendiums will assist in educating carers on the management of PKU on a day-to-day basis, the PKU diet, meal plans, blood spot testing guide, resources list and other useful resources and information. The people caring for these late diagnosed adults are generally not family members and have little or no knowledge of PKU and management of PKU. Carers will be able to contact MDDA with any PKU management queries. This service does not replace the excellent care, treatment and advice provided by the metabolic treating team.



## Global Association for PKU (GAP)

Worldwide, approximately 35 babies are born each day at risk of developing severe and irreversible brain damage by the age of one due to phenylketonuria (PKU). PKU can be simply and quickly diagnosed via the “heel prick” test and if treatment begins immediately most people with this condition will live healthy lives. Currently 65% of babies born in the world miss out on this opportunity. GAP aims to create a global platform for PKU advocacy that includes ensuring universal access to newborn screening and diagnosis, treatments and comprehensive care. MDDA President Monique Cooper and Vice President Louise Healy are founding members of GAP.



## Genetic Service Network of Victoria (GSNV)

MDDA attended “Finding Your Voice” in February, on Rare Diseases Day at the Murdoch Children’s Research Institute facilitated by GSNV. This forum brought together stakeholders from support groups, government, research, clinical and industry to engage and interact with the support sector. The day launched RDNOW – the new rare disease flagship to improve access to rare disease testing at the Royal Children’s Hospital Vic. The day also saw the launch of carrierscreening.org.au – a new website providing information about carrier screening and an update from the Mackenzies Mission team. MDDA participated in sessions to further develop strategies on how to be effective advocates to ensure our community have equitable access to treatment, research and clinical trials.



## The Grand Hotel Warrandyte

The Grand Hotel Warrandyte VIC, is the first establishment in Australia to offer specially designed low protein menu options on their everyday menu. This year they extended their services to the first ever Low Protein takeaway menu. The MDDA appreciate The Grand taking on the task of understanding and catering to the needs of our PKU/IEM community. The Grand generously also provide in-kind support and cooking resources for our retreats, host local events and are always keen to help out with awareness initiatives and fundraisers.



## 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 program 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.





LowPro Inspo is MDDA's Low Protein YouTube Channel and a fantastic way for our community to share delicious LowPro meals with each other! Launched at our 2019 Tasmanian Retreat, LowPro Inspo includes cooking videos that open the door to Australian families showcasing the diverse range of diets of those living with protein IEMs. The channel is initially focusing on low protein cooking and recipes, we also envision it to evolve to include other inspirational content from MDDA, our members and affiliated communities.

Our goal for this channel is to bring the community together through the power of cooking and to inspire each other to make delicious low pro food. Whether you are a cooking fanatic, or a novice, there is something there for everyone.

We are continuing to build this YouTube Channel and have uploaded some great videos from people in our community.



During the COVID-19 Pandemic, with everyone at home, we thought a great way to inspire our community would be to hold our first LowPro Inspo LIVE, providing an opportunity to come together in our own kitchens and share an easy and delicious recipe.

MDDA's resident foodie Bianca hosted this live session demonstrating her Cauliflower Fried Rice. Taking place on Zoom and broadcasting live on Facebook this proved to be a wonderful interactive and successful event, with some delicious results. Our goal is to continue to build the LowPro Inspo YouTube Channel, to inspire more members of our community and learn some delicious meals along the way.





# Year at a glance



## July to September

- ✓ 2 graduate students placed for work experience through GSNV/Melbourne University
- ✓ Member SA - Fun Run
- ✓ Member Vic - Facebook birthday fundraiser
- ✓ Member Vic - Brighton Primary School Dads Social club fundraiser
- ✓ Member Vic - Garage Sale



## October to December

- ✓ Tasmanian Retreat, Tamar Valley Resort
- ✓ Visit to a disability home to provide an informative session to staff on PKU resident
- ✓ Member Advantage program launch
- ✓ Transitional handbook/resource developed
- ✓ Lowpro Inspo - YouTube cooking channel launched
- ✓ End of Year state based celebrations
- ✓ Zest program session
- ✓ Visit to Canberra as part of ongoing advocacy with Department of Health and key MP's.
- ✓ WA MDDA Trivia night, "A Night at the Movies", fundraising night
- ✓ PeeKabU bags sent out to stock clinics for newborn IEM babies



## January to March

- ✓ Attended the National Strategic Action Plan for Rare Disease event at Parliament House in Canberra
- ✓ Rare Disease Day celebrations – Feb 29th
- ✓ MDDA attend the Rare Disease Day event held at the Royal Children's Hospital Vic.
- ✓ Member fundraising - "Show us your Stripes" held at Coles store Mango Hill on Rare Disease Day
- ✓ COVID-19 resource added to website
- ✓ School Starter resource developed



## April to June

- ✓ Low Protein take away menu developed at the Grand Warrandyte
- ✓ Special virtual interest chat groups ran
- ✓ Live Lowpro Inspo cooking sessions held
- ✓ 2 Member Facebook fundraisers held.

# Financials

An encouraging sign during this financial year saw an increase in membership renewals. Although FY20 financials show a deficit of \$6.7k, this can be attributed to the effect COVID-19 had on our ability to fundraise, also the addition of expenses due to website upgrades, development of program initiatives and delayed sponsorship payments. MDDA moved to Xero cloud-based accounting software this year which has simplified reporting requirements and improved efficiency. We are pleased to report that 100% of every dollar received from generous donations and fundraising go directly to the IEM community through educational programs, member support initiatives and research projects.

## Summary of financial results

30 June 2020

Income	
Grants	\$230,000
Sponsorships	\$99,500
Donations	\$12,549
Fundraising	\$5,818
Events	\$23,653
Other Revenue & Subsidies	\$50,950
<small>* MDDA are grateful recipients of Government subsidies inc the Cashflow Boost &amp; JobKeeper subsidy.</small>	
<b>Total Income</b>	<b>\$422,470</b>
Expenses	
Operating Costs	\$64,614
Secretariat Expenses	\$205,414
Member Support	\$84,451
Event/Fundraising	\$25,043
<b>Total Expenses</b>	<b>\$429,236</b>
<b>Loss</b>	<b>\$6,735</b>

A full audited financial report is available on request.

Thank you to the following #teamMDDA member driven fundraising initiatives;

- ✓ WA Members MDDA Trivia night "A Night at the Movies"
- ✓ Hayley Pash WA: Facebook fundraiser
- ✓ Pauline Blyth TAS: Facebook fundraiser
- ✓ O'Shea Family QLD: Show us your stripes
- ✓ Hendricks Family VIC: Garage Sale & Brighton Primary School Dad's Social Club
- ✓ Senada Suljagic VIC: Facebook fundraiser
- ✓ Grasso Family SA: City to Bay Fun Run
- ✓ The Grand Hotel Warrandyte VIC: Fundraiser



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.

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

- ✓ Low Pro Pop Up Kitchen & Hamper Drive
- ✓ IEM Family Assist - retreats & remote clinics
- ✓ My Low Pro PAL
- ✓ PKU Pathways - outreach support, mentoring
- ✓ National PKU & IEM Research & Clinical Care Fund



## 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.



## 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.

# 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 VIC

### Secretary

Bianca Albanese, PKU Adult  
NSW

### Health & Wellbeing

Susi Hendricks, PKU Parent VIC

### Executive Officer

Jenny Briant

### Administration Assistant

Kerry Tulloch

### Communications and Marketing

Bella Hendricks

### Patient Pathways Telehealth

### Nurse

Joanne Campbell



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