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MSUD NEWSLETTER

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REUNITING AGAIN: FROM THE PRESIDENT'S DESK

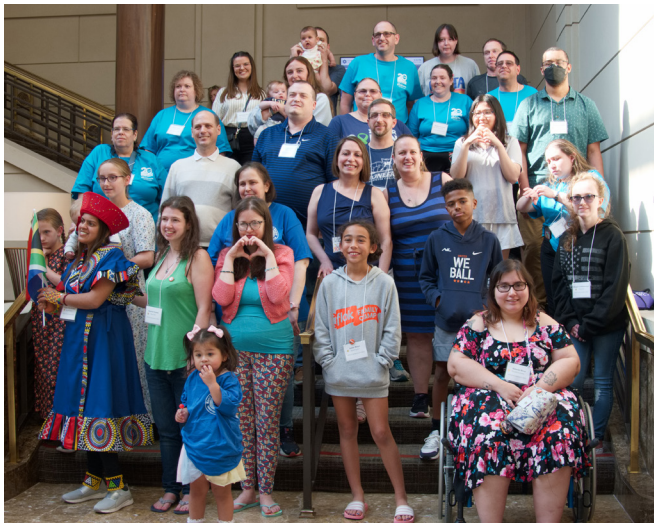
By: Sandy Bulcher, President, MSUD Family Support Group

The 20th MSUD Symposium in 2024 was a huge success! It was wonderful to see so many MSUD friends and to meet new families. In fact, 12 new families attended for the first time, and three of those families had infants with MSUD under one year old. A total of 200 people attended from 20 U.S. states and six other countries. It was exciting to see transplant families present as well. Based on the feedback I received, attendees felt the symposium met their educational and support needs.

The symposium was held from June 27-29, 2024, at the beautiful Hyatt Regency in Reston, VA, approximately 20 miles from Washington, DC. Some families were able to incorporate a vacation while attending the symposium.

In addition to the informative presentations by knowledgeable professionals, families also had the opportunity to interact with vendors and learn about their products and services. Check out the synopsis of a few of the speakers' presentations in this newsletter. To view more of the presentations, visit the MSUD website at www.msud-support.org, where you can find their PowerPoint slides. Ed Fischler, MSUD Vice President, worked tirelessly to record the presentations for the website, but unfortunately, technical issues prevented that from being possible.

We are grateful to the many professionals from Children's National and Georgetown University in Washington, DC, who participated in the 20th MSUD Symposium. In addition, the Children's National metabolic team was available in case anyone with MSUD became ill. Fortunately, their expertise was not needed.



Symposiums like this are not possible without the financial support of many donors. A special shout-out to a few of our top donors, including the United Service Foundation, Marty and Alison Bell, Nutricia, and Vitaflo, for their very generous contributions. If you would like to make a donation in honor of someone with MSUD and support this wonderful event for so many MSUD families, please visit www.msud-support.org/donations-general-fund/.

Hosting a successful symposium requires a great deal of work, but thankfully, I didn't have to do it alone. Our Executive Director, Denise Kolivoski, and I worked closely together on many of the details. Karen Dolins, Sarah Keyrouz, and Taryn Kessel assisted in developing the agenda. Michelle Petty managed the low-protein menu, Zulma Santos coordinated the registration table, and Deb Fall and Whitney Trump supervised the childcare room. Without their help, the event would not have been possible!

During our planning, we heard from some MSUD families who had hoped to attend the symposium but were unable to due to prior commitments. We missed you and hope to see you at the next symposium (time and place to be determined).

As always, feel free to send me an email or call anytime (sandybulcher@gmail.com, 740-972-5619). ■

Inside This Issue:

Sections

FROM THE DIRECTOR'S DEN

20TH MSUD SYMPOSIUM: A CELEBRATION OF FIRSTS

By Denise L. Kolivoski, MBA, Executive Director, MSUD Family Support Group

This year marked the 20th MSUD Symposium, and it was my first time attending this incredible event. The theme of "firsts" was a fitting way to capture the excitement and milestones we all experienced together.

The symposium was held at the beautiful Hyatt Regency in Reston, Virginia, situated in the lively Reston Town Center. With over 30 restaurants and shops within walking distance and a green space just steps outside the symposium venue, attendees had the perfect setting to enjoy some fresh air between sessions.

One of my personal highlights was meeting with the hotel's chef and our dedicated volunteer, Michelle Petty, to plan MSUD-friendly meals. They did an outstanding job, and I even learned about the preparation of low-protein foods like eggs and pasta.

Another first was the remarkable number of MSUD children, individuals, and families I had the pleasure of meeting—all in one place! We had about 200 people in attendance, including families and professionals from over 20 U.S. states and six other countries. It was particularly special to see 40 MSUD individuals, ranging from six months old to 55 years, come together. Among them were eight people who had undergone liver transplants and more than ten families attending their first symposium.

This year, we also had our first presenting sponsor, Nutricia, and the largest number of exhibitors ever. They provided essential samples, education, and resources specifically tailored for the MSUD community.

Meeting so many friendly faces and pioneers in the MSUD community, such as Wayne and Joyce Brubacher,

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FROM THE EDITOR'S LAPTOP

JUNE: THE MONTH OF HIGH LEVELS

By: Susan Needleman, Newsletter Editor, MSUD Family Support Group

For many of you, June 2024 will be a month you remember for attending the 20th MSUD Symposium and meeting others with MSUD in person. For me, it will be remembered very differently. On June 3rd of this year, I ate some bad food, either spoiled or too old, and ended up vomiting. Fortunately, I managed to keep down the formula I had consumed earlier in the day, but as anyone with MSUD can understand, this was frightening—and unfortunately, it was only the beginning.

Over the next week, I believe my levels doubled, which was not surprising since I was not eating much. Even though I was eating low-protein, high-calorie medical foods, it was not enough to meet my daily calorie needs, and my levels kept rising until they were five to six times higher than they should have been. This led to experiencing several panic attacks, which I rarely have, but which happen when my levels get very high.

Some days in June were harder than others. I was only managing to eat about a third to half of my sick-day diet because I had a very little appetite. However, I did begin having an extra batch of formula most days, which provided just enough calories to keep me out of the hospital—although I came very close to having to go. I did not add extra MSUD powder to my regular amount of formula, as some people do, because it changes the taste so much that I have never been able to tolerate it that way. I also, automatically woke up around every 1-2 hours when I did sleep, to drink at least a little formula or Gatorade. I like to think my body woke me up because it knew it needed it. As I continued to drink more formula, I gradually started regaining my appetite, but not for my usual foods. The regular low-protein pasta shapes I typically eat were too large for the little appetite I had.

It was not until near the end of June that I remembered something from my past illness episodes (which last happened so long ago that I had forgotten): I used to eat smaller shaped pasta, easier, when I was sick, and that worked again. I also began to eat low-protein medical pastry products, which provided 150+ calories per serving. I highly recommend keeping these types of

'Editor's Laptop' continued on page 8

TRANSPLANTATION FOR MSUD:
LESSONS FROM THE FIRST 100+ CASES

By: George V. Mazariegos, UPMC Children's Hospital of Pittsburgh

Extensive experience with liver transplantation for MSUD has been gained at our center in collaboration with the Clinic for Special Children as well as the Medical Genetics and Hepatology teams at the Children's Hospital of Pittsburgh. A summary of the presentation at the biennial symposium 2024 follows and the full presentation is available upon request.

Building on the demonstration that liver transplantation cured MSUD in a patient receiving a liver transplant for a separate indication, an elective protocol was developed to safely consider liver transplantation for patients suffering from classical MSUD. [1] In addition to a foundational expectation of surgical and medical transplant expertise at a center of excellence, other critical programmatic components included development of a protocol for medical management of hyperleucinosis in the peri-operative period should unanticipated complications arise, the 24-hour availability of MSUD total parenteral nutrition for patients who might not be able to tolerate enteral feeding and immediate turnaround of amino acid measurements [2]. In the over 100 cases that have followed, we have demonstrated long term 98.4% survival with one retransplant and one patient relisted for transplant. Long term metabolic control and freedom from hyperleucinosis post-transplant with a normal unrestricted protein intake has been documented by our center [3] as well as by sites across the world.[4]

Numerous resources exist to help families in navigating the choice and selection of a transplant center. Family and patient support groups like this MSUD Support Group https://msud-support.org/ and Transplant Families www.transplantfamilies.org are logical first steps. United States program data can be found at www.srtr.org. A collaborative learning network for excellence in transplantation www.starzlnetwork.org was established to facilitate transparency among select institutions committed to urgently improving outcomes in transplantation and also to developing resources that could be shared across all centers https://starzlnetwork.org/resources-landing/.

Transplant options for the child or adult with MSUD include deceased donor or living donor transplants preferably from non-related donors if possible. Although living related transplantation may be necessary and be successful in global regions where deceased donation is not consistently available [5], post-transplant hyperleucinosis crisis has been reported in a child who received a living related graft from her MSUD carrier mother. [6] A wonderful global development in the evolution of transplantation for MSUD has been the ability to utilize the explanted liver from the MSUD patient to save the life of other patients with liver diseases from other causes in what is known as a domino transplant. [7] Domino transplant recipients of MSUD grafts do not develop MSUD nor have any protein restriction.[8]

Critical to success post transplantation is a commitment to long term follow-up both to monitor long term graft health and optimize long term immunosuppression but also to monitor for complications related to immunosuppression including malignancy whose risks, particularly for skin cancers may be increased after solid organ transplantation.[9] I look forward to being a resource to families considering transplantation and can be reached at George.mazariegos@chp.edu.

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NEW CANPKU+ CO-CHAIR IN ATTENDANCE AT 20TH BIENNIAL SYMPOSIUM

By Stacy Mason, Canpku+ Co-Chair, 41 Years Old, Classic MSUD, Warman, Saskatchewan, Canada

My name is Stacy Mason, I am 41 years old and live in Warman, Saskatchewan, Canada, where I am an Educational Assistant, assisting a classroom teacher with students who need extra support. I, also, have classic MSUD. Recently, I became Co-Chair of CanPKU+, the Canadian organization for MSUD and similar rare disorders. I am excited to tell you in future issues of this newsletter, projects that I am working on in this role, with MSUD Chair Susan Needleman.

For now, I want to tell you about my experience attending the 20th anniversary of the biennial MSUD Symposium. The 20th MSUD Symposium was held at the beautiful Hyatt Regency in Reston, Virginia, from June 27 – 29, 2024. The hotel and surrounding grounds were absolutely stunning, complete with a green space out back that featured local music acts on weekends. I had the honor of being one of two Canadian families in attendance, along with my husband, Jon Mason, and my mother, Dianne Quintal, thanks to the help of the MSUD Family Support Group. It was also great to meet up with the other family from Canada. It was remarkable to see MSUD represented at all stages of life, and there was a strong sense of community and belonging among everyone who attended.

As someone on a low-protein diet, I can say the food this year was fantastic! I really appreciate the effort to match the low-protein meals to the regular food. As a person with classic MSUD, I always enjoy when the meals look similar to what everyone else is eating—it makes me feel more included, and it's just easier that way. The hotel provided a pizza dinner on Thursday, breakfast and lunch on Friday, and breakfast on Saturday. Given that they created two separate buffets for each meal, it was clear a lot of work and preparation went into everything.

For Friday night's dinner, we were given a map of the surrounding restaurants, complete with a list of which ones offered the best options for those with MSUD. As you all know, going out for dinner can be tricky for us, so knowing they had pre-checked all the nearby restaurants made for a stress-free meal where I was able to eat a full dinner. Afterward, we wandered around the market square, admiring the beautiful flowers and the colorful light-up fountains. It was truly a beautiful location for the symposium.

The presentations this year were highly informative, with a focus on liver transplants, including one titled "Lessons from the First 100" and another expert presentation, "Georgetown Experience in Liver Transplantation." It was incredible to hear how far liver transplants have come. I also learned about domino transplants, where the original liver is given to someone else, and that person does not develop MSUD. This seems like a promising option for those considering a liver transplant.

Another fascinating presentation covered the progress being made in gene therapy. I'll admit that a lot of the technical details went over my head, but they did a great job explaining the different gene mutations and how MSUD is passed down. The visual aids were particularly helpful in understanding how my body ended up with this disease, and moving forward, it will help me explain MSUD more effectively to others.

There were also several presentations on managing MSUD, including discussions on diet, exercise, and different ways to take our formula. A few vendor tables were set up, from medical food and formula companies, to flok, who is coming out with an app to help manage our MSUD care. The Coordination of Rare Diseases at Sanford (CoRDS) gave a presentation and had a vendor table, as well, encouraging people to register with them, as this helps with MSUD research. I highly encourage anyone who hasn't registered with CoRDS to do so at https://msud-support.org/patient-registry/.

The last session was especially important to me, as it addressed a topic I've been thinking about a lot lately—MSUD and aging. This session included a Q&A panel and breakout group discussions. It was comforting to hear about the struggles and triumphs others in the MSUD community face and know that many of us are going through similar experiences. The support and acceptance in the room were palpable.

Overall, the symposium was an amazing experience, and I'm so grateful to have been part of it. My two favorite moments were listening to everyone's stories and questions—that's where I felt I gained the most knowledge—and seeing the slideshow of pictures from past symposiums. It was incredible to see how much the MSUD community has grown over the years.

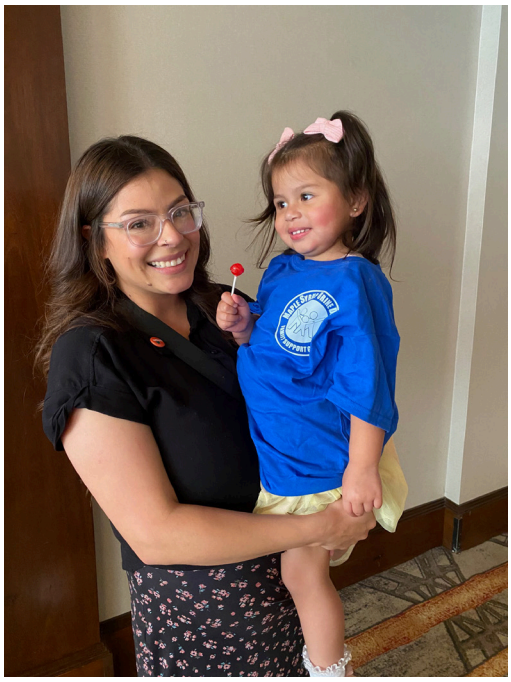
Thank you again to the MSUD Family Support Group for sponsoring us to attend this year's symposium.

Here's to another 20 years!

If you are a Canadian and would like to connect with others with MSUD, attend a Canadian event for MSUD and similar disorders, or just know what resources are available to you as a Canadian, you can contact me at stacylmason31@gmail.com or the MSUD Chair, Susan Needleman at msud@canpku.org or visit http://www.canpku.org. ■

OUR FIRST MSUD SYMPOSIUM

By: Vanessa Anguiano and Jesus Anguiano, Parents of Emma Anguiano, 3 Years Old, Classic MSUD, Southern California, United States of America



We are Emma's parents. Emma has classic MSUD, and she is our enthusiastic, outgoing, and brave 3-year-old. Like most parents, it was really difficult for us to understand Emma's condition at first. But over time, with the support of many MSUD caregivers who shared their experiences and the guidance of our specialist team, we have learned to thrive. We were so fortunate to be able to attend this year's symposium, and it far exceeded any expectations we had.

For starters, it was our first time flying with our g-tube-dependent MSUD toddler, which was an adventure in itself. We landed in Dulles, Washington, filled with excitement. On our first day, we went to register and met so many amazing people who shared their experiences of living with MSUD as adults. We also had the chance to connect with other parents who have gone through similar experiences, which gave us hope and reminded us that we are not alone. We received so many tips on new low-protein foods to try at home, and everyone greeted Emma with such kindness.

Each day, we heard from knowledgeable individuals and doctors who provided us with so much information we hadn't known before. It was incredibly exciting to hear about the advancements in gene therapy directly from the people working on it. We also had the opportunity to speak with and ask questions to people from all over the world, learning how they manage their day-to-day lives with MSUD.

It was inspiring and hopeful to see MSUD individuals with children, some who have undergone liver transplants, and others who attend college and have the opportunity to write their own stories despite their condition. We left the symposium with lifelong friends and immense gratitude for everything that the MSUD Board of Directors has done, to give us all the chance to connect, build our community, and find our people. ■

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was another first for me. The warmth and welcome I received from everyone made the experience unforgettable.

It was also my first year serving as the Executive Director of the MSUD Family Support Group, and I was honored to present to the MSUD community in person for the first time. My presentation focused on the fun in fundraising and the immense good that comes from giving back. I discussed several ways to support the MSUD Family Support Group, including:

- Vehicle Donations: An easy way to contribute to our cause. More details can be found at www.ncsvehicledonations.com/nonprofits/msud-family-support-group/.
- Annual Gifts: A suggested donation of $50 or more helps us continue our work as the only international MSUD organization. Donations can be made at www.msud-support.org/donations-general-fund/ or by sending a check to Dave Bulcher, Treasurer, at 958 Medinah Terrace, Columbus, OH 43235.
- Fundraising Events: Consider organizing a run, walk, or event with your family to benefit our support group.
- Legacy Gifts: Leaving a gift through your will is a powerful way to ensure the future of the MSUD community. I'm here to help guide you through this process.

It would be my pleasure to work with you to fulfill your passion for giving and to create a stronger future for the MSUD community. Please feel free to reach out to me at execdirector@msud-support.org.

As I reflect on my first symposium and my first year as Executive Director, I am filled with gratitude and inspiration. The connections made, the knowledge shared, and the generosity of the MSUD community are what make our work possible. Together, we can continue to advocate for an improved quality of life for all those affected by MSUD. ■

MAKE A DIFFERENCE FOR THE MSUD COMMUNITY!

The MSUD Family Support Group kindly requests a ***suggested annual donation of $50*** to support our mission of education, research, and advocacy to meet the current and future needs of family members, friends, and neighbors impacted by MSUD worldwide. While we have made significant progress in advancing this mission over the past several years, much more remains to be done to improve the quality of life for those living with the daily challenges of MSUD.

Your $50 annual contribution supports your continued involvement with the MSUD Family Support Group, including receiving newsletters like this one, filled with valuable information, and other essential communications. Your ongoing support is greatly appreciated as we work together to further our shared mission. **On behalf of the individuals and families of the MSUD Family Support Group, we encourage you to make a $50 gift today!**

You can show your support online at www.msud-support.org/donations-2. or send a check to:
MSUD Family Support Group
c/o Dave Bulcher
958 Medinah Terrace
Columbus, Ohio 43235 ■

ANNUAL DONATION REQUEST:

The MSUD Family Support Group kindly requests a suggested annual donation of $50 to support the mission of education, research, and advocacy to meet the current and future needs of our family members, friends, and neighbors impacted by MSUD throughout the world. While we've worked hard to move this mission forward over the past several years, much more work is needed to improve the quality of life for people living with the daily challenges of MSUD.

Your $50 annual contribution helps support your continued journey with the MSUD Family Support Group including newsletters such as this one filled with valuable information and other essential communications. Your ongoing support is greatly appreciated as we work together to further our shared mission. **On behalf of the individuals and families who we support, please contribute a $50 gift today!**

You can show your support online at https://msud-support.org/donations-2
or send a check to: Dave Bulcher, MSUD Family Support Group 958 Medinah Terrace, Columbus, OH 43235.

Please complete the information below. Donations support organizational costs that enable us to provide education and support to those affected by the disorder and their families. Your tax-deductible gift makes a difference!

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I/we would like my/our gift to be given: ☐ In honor of ☐ In memory of _____

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☐ I/we have included MSUD Family Support Group in my/our will, trust, or other estate plans.

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Or go to the MSUD website www.msud-support.org to donate and sign up to receive mailings.

20TH MSUD SYMPOSIUM

ADULT BREAK-AWAY SESSION WITH DR. GOLD

By: Nikolai Rudd, MSUD Newsletter Writer, 50 Years Old, Transplanted, North Adams, Massachusetts

On the last morning of this year's MSUD Symposium in Reston, Virginia, over twenty MSUD and MSUD Transplanted Adults gathered with Dr. Gold for a "break-out" session to discuss specific health issues and challenges that have emerged and affected our metabolic levels and overall health. From my personal experience—dealing with thyroid polyps that required biopsies for cancer, low testosterone levels, a kidney stone, and a small stroke possibly linked to new medications—to learning the night before about Jesse Kiel's passing and Libby Stone requiring a second liver transplant after many years, there were a lot of emotions in the room. Women also shared their own body-related challenges.

We also discussed the anxiety issues many of us have faced, either starting during the pandemic or being exacerbated by it. Additionally, several liver-transplanted patients noted difficulties with sleep regulation after their transplants. Issues ranged from insomnia and panic attacks to trouble consistently getting a good night's sleep.

There were so many topics covered that it would be impossible to recall them all, but these were some of the key points that came to mind. For many years, adult MSUD patients felt like we were being left behind. It seemed like doctors learned how to treat younger MSUD patients from the older generation but then shifted their focus to newborns and children. Meanwhile, many of those children were transitioning into adulthood, and now the first and second generations of MSUD patients are entering their forties and fifties, facing typical health issues for these age ranges. It seemed like no one was prepared for how these normal aging challenges could impact metabolic levels until it was too late.

Dr. Gold's study on adult MSUD patients, both transplanted and non-transplanted, has given us a renewed sense of hope. For the first time in a long while, we feel like the medical community is genuinely trying to understand what's happening to us as we age and how it affects our metabolic levels.

This break-out session was incredibly helpful. In earlier symposiums, we had more frequent break-out sessions, which allowed us to connect better with each other. More importantly, these sessions gave MSUD doctors the chance to hear firsthand how stress, jobs, school, diet mismanagement, and other health issues impacted our levels and our lifestyles. While this year's session was shorter than we would have liked, it was still beneficial. I hope that what was learned during this session and throughout the symposium will help improve patient care and strengthen our community.

Lectures provide valuable information, but they don't always help patients understand what's happening with them individually at that moment. Break-out sessions, on the other hand, served a great purpose in the past and should be revived to help all generations of MSUD and MSUD-transplanted patients, as well as the doctors who treat them, to learn and grow together. ■

'Editor's Laptop' continued from page 2

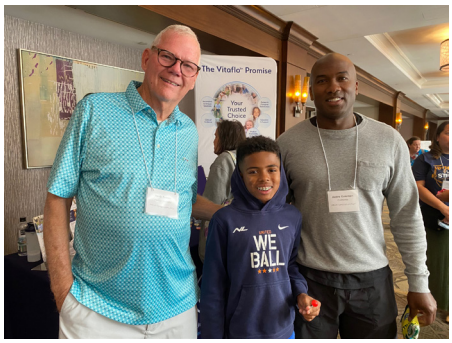
medical foods in the house. Even if you do not typically keep medical foods on hand, or if you cannot get them, it is essential to have something low-protein with a high calorie count. After this episode, I cannot emphasize this enough. While these foods may not have the vitamins and minerals you get from a healthy diet, when you are sick, they are the best options for someone with MSUD to recover. The extra formula compensates for the vitamins and minerals you are missing from your usual diet.

Finally, my leucine levels returned to normal after a month, and two weeks after that, my isoleucine and valine levels did as well. As my levels normalized, my panic attacks subsided too. As I write this in mid-September, I still have not fully regained my muscle tone, but I no longer have brain fog or panic attacks. I cannot help but wish we had more treatments available to counteract the panic attacks that are so common in those of us with MSUD. I cannot help but wonder if there is something—maybe a vitamin, mineral, or other amino acid—that could be added to our formula to help. Regular panic attack medication did not completely eliminate them but did help to some extent.

This is why it is so important to support the MSUD Family Support Group and the research they fund to find better treatments and cures for MSUD. To donate, visit https://msud-support.org/donations-2/.

On another note, I would like to acknowledge Nikolai Rudd, who has transitioned from Assistant Editor to Writer on the Newsletter team. We look forward to reading his articles in this and future publications. ■

MSUD SYMPOSIUM PHOTOS



20 YEARS
BIENNIAL 2024
SYMPOSIUM
MSUD FAMILY SUPPORT GROUP
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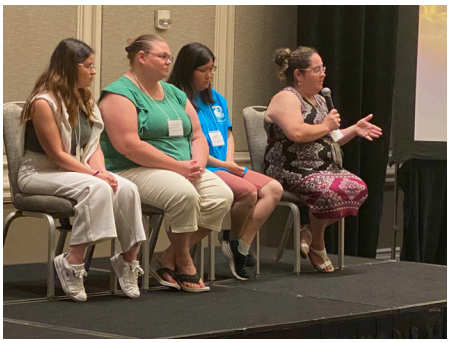

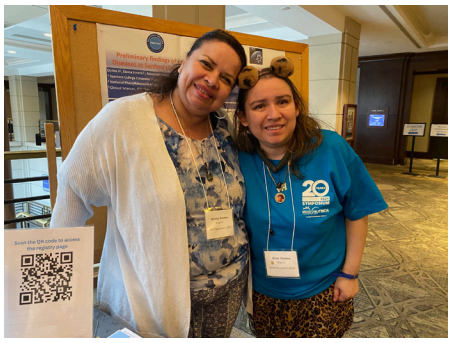

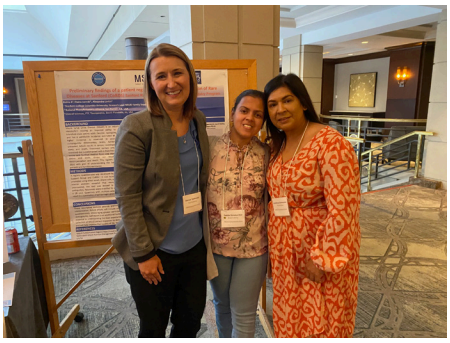

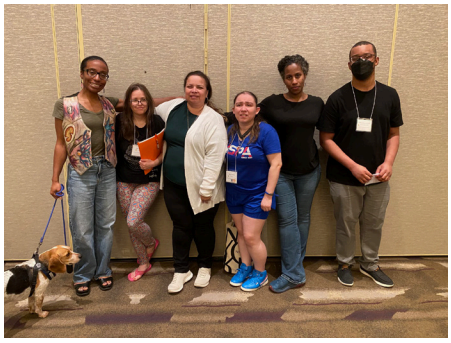

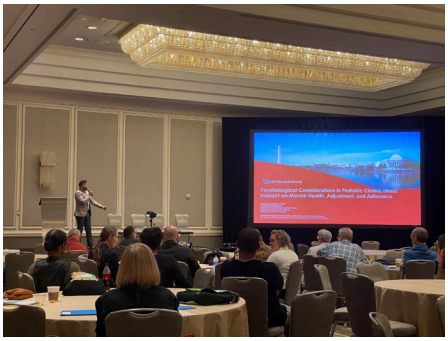

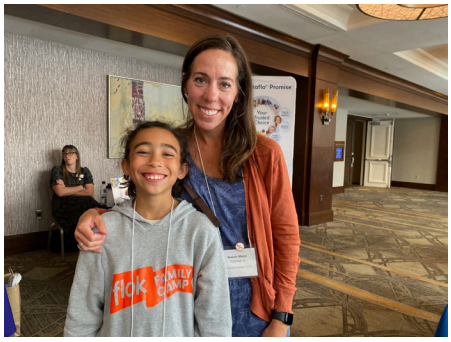

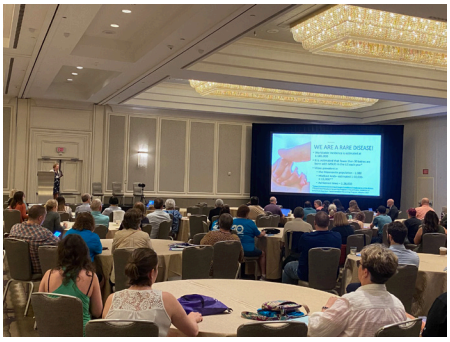

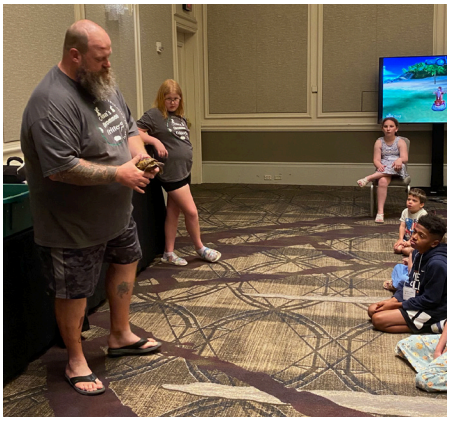

THE ROLE OF ISOLEUCINE, VALINE AND ALANINE IN ACUTE MANAGEMENT OF MAPLE SYRUP URINE DISEASE (MSUD)

Taelyr Mellor, MS, RD, LDN; Susan McGowan, MHS, RD, LDN, CNSC; Shawn Carro, MS, RD, LDN; Abigail Lauko MS, RD, CSP, LDN; Can Ficicioglu, MD, PhD

It is well known that isoleucine and valine are required supplements in the treatment of MSUD. It is also understood that these supplements are often needed in greater amounts during crisis. However, there are no clear guidelines on the dosing of these supplements during a decompensation. Additionally, alanine supplementation is not currently considered standard of care for treating maple syrup urine disease (MSUD), but it has been described as a potentially beneficial approach by several experts in MSUD management (Morton et al1, Nyham et al3, and Strauss et al4). Alanine is usually lower than normal especially during acute metabolic crisis. There are a few key reasons for this:

1. In MSUD, there is an accumulation of branched-chain ketoacids (BCKA) due to the deficiency in branched-chain α-ketoacid dehydrogenase complex. This leads to increased transamination reactions that favor the formation of branched-chain amino acids (BCAAs).
   BCKA + Glutamate == BCAA + Alpha-ketoglutarate (AKG)

The transamination reactions that produce BCAAs consume glutamate as an amino group donor. This depletes glutamate levels. Alanine is normally produced from pyruvate and glutamate via the enzyme alanine aminotransferase. With glutamate levels depleted, there is less substrate available to produce alanine, making alanine conditionally essential during crisis.2,4
Glutamate + Pyruvate == Alanine + AKG

2. During acute metabolic crises in MSUD, there is increased protein catabolism and breakdown of muscle tissue. This further reduces alanine levels, as alanine is an important amino acid released from muscle during catabolic states to produce glucose in the liver. Alanine is a major gluconeogenic precursor. It is formed primarily in skeletal muscle and then transported to liver (and kidney), where it can be converted to glucose.5

3. The accumulation of toxic metabolites in MSUD can impair normal amino acid metabolism and transport, potentially further reducing alanine production and levels.

4. Low alanine levels are often used as a marker of catabolism in MSUD and other metabolic disorders.

Why Alanine supplementation is important especially during metabolic crisis: Alanine promotes anabolism and can prevent catabolism - key goals in managing acute MSUD decompensation. Management of decompensation usually includes dextrose fluids, intravenous lipids, enteral metabolic formulas, and or, if accessible, a parenteral amino acid solution free of branch-chain amino acids. These interventions are provided alongside supplements of isoleucine and valine. The end goal is to shuttle leucine from the plasma into protein synthesis and help prevent more leucine from entering plasma with muscle breakdown. Supplemental alanine could help prevent skeletal muscle breakdown by providing an alternative substrate for gluconeogenesis and spare energy requirements by turning off the alanine-glucose cycle.3, 6 Monitoring alanine levels can provide insight into the metabolic state of MSUD patients.

During well-state, isoleucine and valine are supplemented to prevent deficiency on a protein restricted diet with an attempt to maintain physiologic ratios. Valine is ideal close to double the leucine level and isoleucine about one-to-one given the relative affinities for transport into the brain.3,4 This addresses one reason we desire to increase levels of valine and isoleucine relative to leucine with supplementation during encephalopathic crisis.

Valine and isoleucine also promote protein synthesis. When isoleucine and valine are depleted relative to leucine, they become rate-limiting for protein synthesis and the removal of leucine from plasma slows or stalls completely. With high dose provision of isoleucine, valine, and other BCAA-free amino acids we are ensuring swift removal of leucine from plasma. Morton et al1 and Strauss et al4 describe ideal supplementation of valine and isoleucine during crisis to be between 20-120 mg/kg and recommend maintaining plasma valine and isoleucine levels between 400 to 800 umol/L.1,4 This represents a wide range of recommended dosing and a lack of guidance for use of the higher versus lower end of this range. It is also unclear where to start with supplementation during initial presentation or when BCAA levels are not known in a child or adult presenting symptomatically.

Using available literature on isoleucine and valine supplementation, and our experiences with the addition of alanine supplementation, Children's Hospital of Philadelphia will develop protocols that have measurable

outcomes pre- and post-implementation. The protocols will provide dosing guidance for isoleucine and valine based on weight and leucine levels. Guidance regarding alanine supplementation will also be provided. While not standard practice, alanine supplementation represents an intriguing approach to MSUD management based on its metabolic roles. It could complement other anabolic strategies like dextrose fluids, lipids, and BCAA-free amino acid solutions. However, further clinical research is needed to definitively establish its efficacy and optimal use in MSUD treatment protocols.
References:

1. Morton DH, Strauss KA, Robinson DL, Puffenberger EG, Kelley RI. Diagnosis and treatment of maple syrup disease: a study of 36 patients. Pediatrics. 2002;109(6):999-1008. doi:10.1542/peds.109.6.999
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3. Nyhan WL, Rice-Kelts M, Klein JD, Barshop BA. Treatment of the acute crisis in maple syrup urine disease. Archives of Pediatrics & Adolescent Medicine. 1998;152(6). doi:10.1001/archpedi.152.6.593
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5. Goday PS, Mehta N. Pediatric Critical Care nutrition. McGraw Hill Professional; 2014.
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MSUD AND EMERGENCY LETTERS

Jessica Scott Schwoerer, M.D., Associate Professor, Dept. of Pediatrics - Genetics Medical College of Wisconsin, Children's Wisconsin Metabolic Consultant, Wisconsin Newborn Screening

When I see patients with metabolic disorders like MSUD, I focus on helping them prevent and prepare for injury or illness by emphasizing several important steps. These include ensuring that they are up to date on immunizations, practicing good hand hygiene, following a sick day diet, having emergency letters ready, and wearing a medical bracelet.

Illness is a part of life, but for a patient with MSUD and their family, it not only brings the misery of sickness but also the stress of managing a sick day diet, overnight feeds, emergency amino acid level checks, and the potential for hospitalization. Injury is also inevitable. Whether it's a fall from the monkey bars, an unexpected hit to the head, or something more severe like a car accident, the risk of metabolic decompensation during illness or injury is real.

There are steps you can take to be prepared in case of injury or illness. It's essential to have an emergency letter on you, from your metabolic team, ready to give to any healthcare provider, particularly in an emergency department. This letter should include:

- Your name, date of birth, and MSUD diagnosis
- A summary of MSUD, including risk factors for decompensation and signs/symptoms of metabolic decompensation
- Initial steps for the emergency department, including lab tests and starting intravenous (IV) fluids
- Contact information for your metabolic team, both during the day and for after-hours emergencies
- Resources such as the GeneReviews website or the New England Consortium of Metabolic Programs (https://www.newenglandconsortium.org) for providers to better understand MSUD and your care

This letter helps initiate evaluation and management when you arrive at the emergency department, while also ensuring that current care providers know how to contact your metabolic team. You can keep your emergency letter with your formula kit, in a purse or backpack, or even take a picture of it on your phone for easy access.

In addition to an emergency letter, MSUD patients should consider getting a medical bracelet. As individuals with MSUD gain independence and are not always accompanied by a caregiver, a medical bracelet becomes increasingly important. It can provide crucial information to emergency care providers and the emergency department, enabling timely and accurate care.

As you review your MSUD care checklist, make sure that emergency letters and medical bracelets are two key items every person with MSUD has.

For more information, visit:

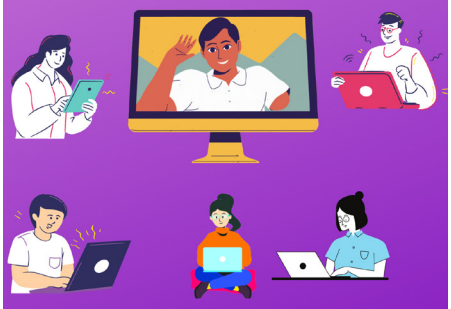
- GeneReviews® - NCBI Bookshelf (nih.gov): https://www.ncbi.nlm.nih.gov/books/NBK1116/
- New England Consortium of Metabolic Programs: https://www.newenglandconsortium.org/
- Medical Alert Foundation: https://www.medicalert.org/ ■

MSUD Teen/Adult

Virtual Meet-Up

Online Occasional Saturday Nights at 8PM EST

RSVP:
susanneedleman.msud@gmail.com



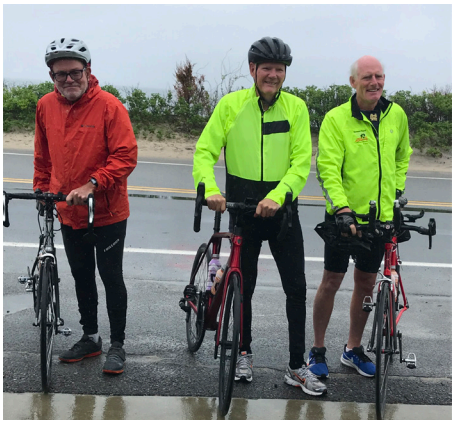
NEED A CHAT?

JOIN OUR ZOOM CAFE
MORE INFORMATION
AT
WWW.CANPKU.ORG/

CanPKU+ offers online cafes where you can join folks like you. Chat about the ups and downs or just the day to day!

ANOTHER SUCCESSFUL BIKE RIDE

MSUD Board member, Herb Foster, held another successful fundraiser bike ride this year, raising $22,000 for Team Scott, in memory of his late son, who had MSUD. Every year, Herb rides one mile for each year of his age; this year, he rode 76 miles. Every dollar he raises goes to the MSUD Family Support Group. ■



DISHING UP HOPE FOR THE MSUD COMMUNITY!

Looking for a fun and delicious way to support the MSUD Family Support Group? Partner with GroupRaise to host a restaurant fundraiser! With over 10,000 participating restaurants, it's easy to organize a group meal where a portion of the proceeds goes directly to support our cause.

Simply request a date at a local restaurant, spread the word on social media, and enjoy a meal with friends and family. The restaurant will donate a percentage of sales back to help make a difference in the lives of those with MSUD. Let's dine for a cause and raise funds to support our community! If you and your friends and family participate, please send us a photo of your group enjoying the meal! We'd love to share your smiles and help spread the word about how dining together supports the MSUD community. Email Denise L. Kolivoski, MBA, at execdirector@msud-support.org. To start your Dishing Up Hope for the MSUD community event, visit https://www.groupraise.com/. ■

DISHING OUT HOPE FOR THE MSUD FAMILY SUPPORT GROUP

SUPPORTING MSUD INDIVIDUALS AND FAMILIES: DONATE YOUR VEHICLE TODAY!

Are you looking for a meaningful way to make a difference in the lives of individuals and families affected by MSUD? Look no further! The MSUD Family Support Group is excited to offer a new opportunity for support through vehicle donations. By donating your vehicle, you'll help advance our mission of education, support, and advocacy for the MSUD community.

Why choose vehicle donation? It's simple, convenient, and tax-deductible. Plus, it provides an eco-friendly way to dispose of unwanted vehicles while contributing to a worthy cause. Whether it's a car, truck, RV, or boat, your donation makes a real impact!

Here's how it works:

1. Schedule a pickup or drop-off location for your vehicle by visiting this link, https://www.ncsvehicledonations.com/nonprofits/msud-family-support-group/.
2. Complete the necessary paperwork – which is a few easy forms.
3. Receive a tax-deductible receipt for your donation.

To learn more about donating your vehicle to the MSUD Family Support Group, visit https://www.ncsvehicledonations.com/nonprofits/msud-family-support-group/ or contact Denise Kolivoski at execdirector@msud-support.org.

Together, we can drive positive change and support MSUD families on their journey toward a brighter future. Thank you for your generosity and compassion. ■

ANOTHER MILESTONE FOR OUR SUPPORT GROUP

2ND MSUD SCIENCE SUMMIT JUNE 27TH, 2024 RESTON, VA

By Karen Dolins, Research Lead

Let me assure you - scientists are hard at work trying to find better treatments and even a cure for MSUD. This was evident when we brought key researchers together in Reston, Virginia the day before our 2024 Symposium, to discuss their work.

Members of our impressive scientific advisory board were joined by an international cast of scientists:

- Dr. Kevin Strauss updated us on the MSUD cow gene therapy project. Petunia, a cow born with MSUD, received gene therapy developed by scientists at the University of Massachusetts as a newborn. She is still thriving at almost 3 years of age and cared for at the Tufts Veterinary School.
- Dr. John Counsell, University College London, discussed his work investigating strategies for delivering gene therapy to the liver and brain. Learn more at https://www.counsellresearch.com/research-projects
- French researchers Drs. Manuel Schiff and Clement Pontoizeau joined us remotely from Paris to describe their advances using gene therapy in MSUD mice.
- Dr. Brad Pentelute of MIT informed the group of his work using messenger RNA (mRNA) as a therapeutic tool. He hopes to develop this technique to treat MSUD.
- Dr. Lital Alfonta of Ben Gurion University in Israel updated us on her work to develop a home leucine monitor. You can read more about this project in the Spring 2024 issue of our newsletter.
- Dr. Jessica Gold provided data on her research on neurocognitive outcomes and executive functioning in adults with MSUD.
- Dr. Rebecca Ahrens-Nicklas of CHOP discussed her work developing a mouse model to explore the effects of MSUD on the brain.
- Dr. Kosar Khaksari of Children's National described her investigation of brain function in MSUD.
- Dr. Richard "Max" Wynn discussed small molecules and how they may be used to develop therapies for MSUD.
- Dr. George Mazariegos of discussed outcomes and challenges of liver transplantation as a therapy for MSUD.

This forum provided a wonderful opportunity for researchers to learn from one another and foster collaborations. The MSUD Board of Directors and clinicians in attendance were awed, energized, and encouraged by the progress being made. We'll continue to update you in this and future issues of this newsletter! ■

RESEARCHERS STUDY BRAIN ACTIVITY AT THE MSUD SYMPOSIUM

By Karen Dolins, Research Lead

The MSUD Family Support Group has provided funding to support the work of Dr. Andrea Gropman and Dr. Kosar Khaksari of Children's National Hospital in Washington, DC. This project uses a non-invasive technique called Functional Near-Infrared Spectroscopy (fNIRS) to measure brain activity. Dr. Khaksari and research coordinator Alexa Taylor joined us in Reston, Virginia with their device so they could test volunteers on site. Registrants for our symposium were contacted in advance and invited to join the study. The researchers were pleased that 8 people signed up in advance and thrilled that after hearing a description of this work and it's importance a total of 21 individuals (MSUD and controls without MSUD) participated. Stay tuned to learn of their findings! ■

MSUD PATIENT REGISTRY WITH CORDS

By Karen Dolins, Research Lead

Dr. Ben Forred, Director of Translational Research, and the CoRDS Registry, joined MSUD Research Lead Dr. Karen Dolins at our 2024 MSUD Symposium to explain why everyone's participation is critical for the future of MSUD research.

A robust registry will help researchers and clinicians understand the challenges of living with MSUD. It will also help us understand how living with MSUD changes from childhood through the adult years and identify gaps currently existing in MSUD research.

As biotech companies work to develop medicines to help treat MSUD and improve quality of life, they will need to submit applications to the FDA. A robust registry strengthens these applications by demonstrating the unmet needs of our community.

We've said it before, and we'll say it again: **PARTICIPATING IN THE CORDS MSUD REGISTRY IS THE MOST IMPORTANT THING YOU CAN DO RIGHT NOW TO IMPROVE THE LIVES OF THOSE WITH MSUD.**

We need everyone to participate, regardless of the type of MSUD, geographical location, or transplant status. Family members of deceased individuals with MSUD should also complete the questionnaires.

Completing the questionnaires takes about 20-40 minutes and you don't need to complete them in 1 sitting. Most questions are easy to answer, but if you're unsure of how to answer a question and need clarification, PLEASE ASK US!

We now have 200 participants, which is awesome. Unfortunately, not everyone has completed all 3 questionnaires:

1. Registration
2. CoRDS general questionnaire
3. MSUD specific questionnaire

If you start the process, please take the time to complete all 3 questionnaires at your earliest convenience. If you completed them but left some questions blank, please don't hesitate to contact us (see below) for clarification. It is important to update your information annually, but it's simple to do so. Only answer questions where the information has changed.

Start the registration process at https://cords.sanfordresearch.org/activation-form.

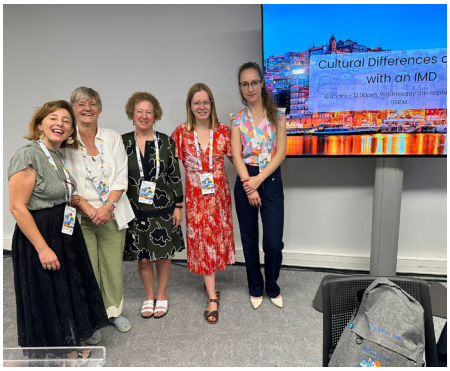
To request assistance, a hard copy of the questionnaires, email or call CoRDS: cords@sanfordhealth.org or 877-658-9192.

If you're unsure how to answer questions, email Karen Dolins at karen.dolins@gmail.com or call her at 914-391-2982. ■

SOCIETY FOR THE STUDY OF INBORN ERRORS OF METABOLISM (SSIEM)

ANNUAL MEETING SEPTEMBER 2-4TH, 2024 PORTO, PORTUGAL

By Karen Dolins, Research Lead



The Society for the Study of Inborn Errors of Metabolism (SSIEM) held its annual meeting in Porto, Portugal, from September 2-6, and MSUD Research Lead, Dr. Karen Dolins, was in attendance. MSUD was featured in several posters and presentations, including exciting developments in gene therapy. Karen participated in a panel discussing cultural differences in managing inborn errors of metabolism and presented on the challenges many individuals around the world face in accessing care and metabolic formula. The meeting provided an excellent opportunity for the MSUD Family Support Group to connect with researchers and industry members, highlighting the need for improved therapies for MSUD. ■

MSUD FAMILY SUPPORT GROUP RESEARCH AGENDA AND UPDATE

By Karen Dolins, Research Lead

The aim of our research agenda is to drive research which will improve the quality of life for those with MSUD. Ultimately, we hope that our research efforts will help:

- Improve treatment
  - o Allow more natural protein in the diet
  - o Prevent a metabolic crisis when ill
  - o Address the impact of MSUD on the brain
- Find a cure for MSUD
- Develop a monitor which will allow families and clinicians to measure blood leucine levels in the home and provide results within minutes
- Understand the needs of those with MSUD as they age

We formed a Scientific Advisory Board in 2020 to help us achieve these goals. Scientists on the board help us develop our research strategy, prioritize projects, and review proposals sent by scientists seeking our support.

The esteemed scientists who sit on our board are:

- Dr. Lindsay Burrage, Baylor College of Medicine
- Dr. Andrea Gropman, Children's National Hospital
- Dr. David Fischler, Xyzagen
- Dr. Irini Manoli, National Institutes of Health
- Dr. Kevin Strauss, Clinic for Special Children
- Dr. Dan Wang, University of Massachusetts
- Dr. Tilla Worgall, Columbia University
- Dr. Richard "Max" Wynn, UT Southwestern Medical Center
- Dr. Joshua Baker, Lurie Children's Hospital

With the help of our generous donors, we have provided support for the following research projects:

- Gene therapy
- Dr. John Counsell
- Dr. Kevin Strauss
- MSUD and the brain
- Dr. Rebecca Ahrens-Nicklas
- Dr. Jessica Gold
- Dr. Andrea Gropman and Kosar Khaksari
- Leucine monitor
- Dr. Lital Alfonta: Ben Gurion University of the Negev
- Dr. Milan Stojanovic: Columbia University

Several companies are working on possible therapeutic agents to help manage MSUD. They include:

- **Zevra:**
  - o Investigating the potential for sodium phenylbutyrate to reduce blood leucine levels.
  - o Previously investigated by Acer Therapeutics.
- **Syntis Bio:**
  - o Developing an enzyme therapy to be administered orally which would break down leucine in the gastrointestinal tract. This would potentially lower blood leucine levels and allow for more natural protein in the diet.
  - o Previously developed by Codexis.
- **Aperiam Bio:**
  - o Developing an enzyme therapy which would be inhaled into the lungs and break down leucine circulating in the blood. This would potentially lower blood leucine levels and allow for more natural protein in the diet.

We share our research aims with investigators in a variety of ways, including exhibiting at professional meetings and through our website. Interested parties are invited to submit a research proposal by emailing research@msud-support.org. We welcome donations of any amount to support MSUD research! https://msud-support.org/donations-research/ ■

CALL FOR RESEARCH PROPOSALS

By Karen Dolins, Research Lead

The MSUD Family Support Group works to improve the lives of individuals with Maple Syrup Urine Disease (MSUD) and pursue a cure. The MSUD Research Fund is a campaign of the MSUD Family Support Group (MSUD FSG) to advance the science of MSUD by funding the most promising research that will lead to new therapy discoveries and a cure.

The goals of the Fund are to:

- Have at least one new treatment therapy in the development pipeline in the next five years.
- Make at least two substantial grants to projects each year that focus on new drug development, new treatments and/or a cure. A variety of treatment solutions need to exist for MSUD patients because it is not a one-size-fits-all disease.
- Fund fellow placements in labs where additional support will speed up the drug development process.
- Fund the most critical projects that have the potential to raise the quality of life of a MSUD patient on a day-to-day basis so they can live the best life possible.

The MSUD-FSG is seeking proposals that will help us meet the above strategic goals as well as to close existing gaps in the knowledge and science of MSUD, and to further support existing MSUD-related projects.

Eligibility of Applicants
Eligible applicants must have a Ph.D., M.D., or equivalent degrees and currently hold a full-time position at any level (post-doctoral, research scientist, assistant/associate/full professor, etc.) at an established academic/research institution. Applicants will be considered from both the United States and internationally. Investigators from centers with a history of research in metabolic diseases are strongly encouraged to apply.

The average grant amount is approximately $25,000. Initial grants are awarded for one year with potential of renewal based on progress made during the first year. Applicants are encouraged to use the funding to support innovative seed or pilot projects to produce preliminary data that would facilitate more substantial funding from federal agencies, foundations or corporations.

Application Guidelines
Applicants are invited to submit a full proposal (single-spaced with 1.0" margins using no less than 11-point Arial fonts). Full proposals should include the following in order and be submitted in one electronic document in the order listed below and be no more than 15 pages in total:

- Project Title
- PI Name
- Abstract/Summary of Project – no more than 350 words
- Introduction
- Hypothesis to be tested
- Significance of proposed research
- Specific aims
- Methods and Procedures
    - o Cells, animals, or patients selection/handling
    - o Research methods/tools
    - o Materials used
    - o Data collection methods and analyses
- Proposed results & potential pitfalls with some solutions
- Project timeline
- Project Budget to include an institutional fiscal officer's signature and itemized justification (no more than one page)
- Project Personnel (no more than 3-page NIH type short bio-sketch).

Applicants without a faculty appointment (e.g., post-doctoral level) must also submit a letter of support from their project mentor. The award requires that funds not be used for the support of institutional indirect or overhead costs.

Timeline
Proposals are considered on a rolling basis and are reviewed by our Scientific Advisory Board. Accepted proposals will be funded by the MSUD Family Support Group for one year. A progress report will be required at 6 months and at 12 months. Awardees may be asked to write a brief article for our newsletter explaining their work in lay terms. A final report will be due 90 days after the end of the funding period.

Please submit proposals and all materials in the order listed and in one document electronically to:

Karen Dolins, EdD, RDN
Research Lead, Maple Syrup Urine Disease Family Support Group
karen.dolins@gmail.com ■

ADVOCACY UPDATE

By Jordann Coleman, Advocacy Chair

In 2024, a crucial piece of legislation is set to impact both the MSUD and rare disease communities. The Medical Nutrition Equity Act (MNEA) stands at the forefront, assuring comprehensive coverage for medically necessary foods when prescribed by a healthcare professional, whether through public or private insurance. The MSUD community, in particular, relies heavily on the passage of this bill, which was originally introduced back in 2018. Previous iterations garnered significant support from various members of Congress (the previous bill has 105 co-sponsors in the House!). The bill was introduced in the House of Representatives as H.R. 6892 this past December. The Senate version of the bill will be introduced later this year.

Your support is pivotal in advancing this crucial legislation. We urge you to share your personal experiences and stories, emphasizing the significance of having coverage for medical nutrition through this bill. Please visit https://nutritionequity.org/share-your-story/ to express why this coverage is essential for you and your family. Your personal narratives hold tremendous weight in garnering legislative backing. Stay tuned for further calls to action to garner support for the Senate & House versions of this bill. Together, we can make a difference. ■

ORGANIZATIONAL AND PROFESSIONAL CONTACTS

This Newsletter does not attempt to provide medical advice for individuals. Consult your specialist before making any changes in treatment.

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WANT TO SHARE YOUR STORY?

Have a story, milestone, or something else you want to submit to our next newsletter?
Email the editors msudeditor@gmail.com or call/text Susan 781-420-2676.

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