MSUD
FAMILY SUPPORT GROUP

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MSUD NEWSLETTER

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FROM THE PRESIDENT'S DESK

By Sandy Bulcher

This winter brought colder temperatures than typical for many of us. As I write this article, it is 6 degrees Fahrenheit outside with 10 or more inches of snow on the ground. Winter leaves me longing for summer with warm temperatures and sunshine. And summer also means that we will be coming together for the 2026 MSUD Symposium, which will be held at the Embassy Suites by Hilton Cleveland- Rockside in Independence, OH on June 25-27th. This symposium has something for everyone affected by MSUD including new, experienced, and transplant families. We've put together an agenda with great speakers and topics. This symposium will include more breakouts, which provide an opportunity for small group discussions.

If you are interested in attending the symposium, you need to register, because we need an accurate count of attendees to determine our food and space needs. You can register by completing and returning the form that was mailed to you or online at www.msud-support.org. You also need to make a hotel reservation by calling the hotel sales coordinator at 216-617-2122. State that you are attending "MSUD 2026 Symposium" or make your reservation online via the MSUD website. The special rate of $145 plus tax for a king suite room and $155 plus tax for a double queen suite room is only available until June 3rd, so don't delay.

If you have questions and/or need support in your MSUD journey, feel free to check out our website - https://msud-support.org/ , webinars - https://msud-support.org/educational-videos-webinars-and-podcasts/, Facebook groups, and phone. If you would like to reach out to a family that has an MSUD child/adult of a specific age, type of MSUD or challenge, I will do my best to connect you with a similar family.

Lastly, many of you receive communication via our e- blasts but if you would also like to be included in our database and receive physical mailings, please call or email me with your name, mailing address, email and phone number and the MSUD individual's name, birth date and type of MSUD and I will include you in the database. Reminder: if your mailing address, email or phone number changes, please update me as well so that I can keep our database as current as possible.

I can be reached at 740-972-5619 or email at sandybulcher@gmail.com.

See you in Cleveland in June!

Inside This Issue:

Sections

FROM THE DIRECTOR’S DEN

By: Denise L. Kolivoski, MBA
MSUD Executive Director

Spring is a season of growth and forward momentum, and I wanted to take a moment to reflect on what Rare Disease Day makes possible when our community comes together.

One of the most meaningful parts of this season is hearing directly from families who choose to step forward and share their experience. I was grateful to connect with Kathy So, who reached out to share a photo of her daughter, Ziva, and a family photo in stripes. Kathy wrote with the kind of generosity that defines this community—wanting to help “in any way that we can,” and continuing to share Ziva’s journey with honesty, including both the ups and the hard days. Their message, shared from Connecticut, is a powerful example of what it looks like to turn lived experience into awareness and connection.

If you’ve ever wondered, “How can I help?” this is your invitation. The MSUD community needs more MSUD Ambassadors—people who are willing to lend their voice, their time, or their skills to help us expand our advocacy outreach and strengthen support for families. Ambassadors can participate in ways that fit their comfort level and availability, whether that’s sharing personal experiences (or helping others share theirs), engaging with advocacy alerts, meeting with policymakers, writing letters, or simply helping amplify our messages when it matters most.

Advocacy is only one area where your involvement can make a real difference. We also need community members who want to support outreach and awareness, welcome and connect with newly diagnosed families, assist with fundraising efforts, help promote educational webinars and events, contribute professional expertise, support projects like the patient registry, or even help with practical needs like content review, translations, and resource sharing. Every role matters, and there is no “too small” contribution when it helps a family feel less alone—or helps move the field forward.

I hope Kathy and Ziva’s example inspires you to reach out to me so we can talk about how we can work together to make MSUD more visible, strengthen support for families, and advance the priorities that matter most to our community. If you have an area of interest—advocacy, outreach, peer support, fundraising, education, or something uniquely your own—please email me at execdirector@msud-support.org. Let’s find the right fit for you, and let’s build what comes next—together.

32nd Annual Metabolic Camp

32nd Annual Metabolic Camp
June 8th - 13th 2026

Emory University | Atlanta, GA
Rani H. Singh, PhD, RDN, LD, *Camp Director*

Contact Us!
Email: **metcamp@emory.edu**
Phone: **(404) 778-8521**
Website: **https://metcamp.net**

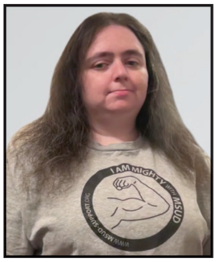
The Annual Metabolic Camp at Emory University (http://metcamp.net) was developed by Rani H. Singh, PhD, RDN, LD to provide a supportive environment for girls and women with PKU and MSUD. A holistic approach to managing these disorders is emphasized, with particular focus on nutrition, genetics, and overall well-being, with the intention of arresting the disease process and minimizing the instances of miscarriages and severe birth defects which are high in this population. We plan to have approx. 30-40 attendees for camp, most are in junior high/high school but ages range between 12 yrs.-adult. We receive limited funding to operate the camp each year--in the form of grants, registration fees, and donations. Watch our Metabolic Camp 2026 promotional video.

Recruiting Campers
The overnight in-person Metabolic Camp will take place on June 8-13, 2026. We would love to have new campers join us, as well as returning campers! Partial scholarships are available for patients who need financial assistance to pay the $375 camp registration fee.

Registration Deadline is April 30, 2026. Contact the Metabolic Camp Staff at metcamp@emory.edu or call 404-778- 8521 to register.

FROM THE EDITOR'S LAPTOP

By: Susan Needleman
MSUD Newsletter Editor



Seize the day! Now more than ever (at least I feel like) I am hearing people asking about when the next treatment of MSUD is going to be out and when will more be known about MSUD. While these answers are on the horizon I wanted to take a moment and reflect on what we have and (hopefully) put into perspective how far we have come.

I was born in 1990, which was a very different time in the MSUD community. I can say I am lucky to have been born in a place that tested for MSUD during newborn screening because a lot of states in the United States (U.S) still did not back then. I had to have dialysis when I was diagnosed, leaving me with scars on my stomach for life. My week-old body already had very high levels leaving me in a metabolic crisis and at the time it was the fastest way to get them down. Growing up my diet consisted of the few medical foods that were sold (most tasted like tapioca) and one of the only three MSUD formulas on the market.

As for support from the MSUD community, there were the symposiums every other year, some families did local events here or there but that was it. If you wanted to learn more about MSUD, from a place other than your clinic or the MSUD community, all that you might be able to find was a line about MSUD in a book at the library. Chances are that line wasn't anything positive about our development.

It was known that MSUD could affect a child's mood and behavior, but little was known about what happened to adults. This was because they were very few adults with it, it had been discovered too recently. Those that were starting to grow into adulthood, had an unknown future, and it was strongly recommended that MSUD women did not get pregnant because it was too dangerous for their MSUD bodies.

Now MSUD is tested in every state in America during Newborn screening. MSUD TPN IV is on the medical market, and most MSUD babies in the United States (U.S) are given it, to lower levels when they are diagnosed and sick patients of all ages. There are currently 16 MSUD formulas available on the market in the U.S, a nice selection of medical foods, and plenty of gluten free foods that happen to also be low in protein, and a plant-based diet is all the rage. It is now known that many MSUD adults get anxiety and panic attacks. MSUD women can now go through pregnancy, stay healthy throughout, and have a healthy baby, live full lives along with MSUD men. If you search the internet today, for "Maple Syrup Urine Disease (MSUD)" you will find countless pages about it, many positive.

Today with the help of the internet and social media, the MSUD community is only a finger tap away. There are Facebook groups and pages, webinars and meet ups, meeting a wide range of supports and needs. For those that do not have the internet there are other ways that they join together to support one another. For those that are in countries that do not have all these resources, they know that there are out there and it is just a matter of how to get it to their country, not how to discover it.

While it is always good to look to the future and new developments, it is important to be grateful for how far we come and appreciate it for that.

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 print and 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!

☐ **$50** ☐ **$100** ☐ **$250** ☐ **$Other** ____________________

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

☐ **Please remove me from the mailing list.**
☐ **Please contact me about making a multi-year pledge and/or estate planning strategies.**
☐ **I/we have included MSUD Family Support Group in my/our will, trust, or other estate plans.**

Name Professional Title (if applicable) Telephone

Mailing Address City State Zip Country E-mail

Families: Spouse's Name MSUD Child's or Children's Name(s) Birth Date(s)

Make checks payable to: ***MSUD Family Support****. Mail completed forms to:*
Dave Bulcher, MSUD Family Support Group Treasurer • 958 Medinah Terrace, Columbus, OH 43235
Or go to the MSUD website www.msud-support.org to donate.

A Law that Could Change Everything for Families Like Yours

By: Jordann Coleman, MSUD Advocacy Chairperson and Board Member

The Medical Foods and Formulas Access Act of 2025 would require insurance to cover the specialized nutrition that keeps your family alive.

If your family lives with MSUD you know that specialized formula or low-protein medical food is not a luxury — it is medicine. And yet, insurance companies routinely refuse to pay for it, leaving families to shoulder costs $1,500 or more per month for formula alone.

The Problem: A Broken Coverage System

Insurers routinely cover costly pharmaceuticals and biologics — drugs that can cost hundreds of times more than medical nutrition. Yet, they deny coverage for physician-prescribed formula. When coverage is granted, insurers often require it be delivered through an invasive feeding tube, even when a patient can consume it orally. State laws offer some protection, but vary widely by age, diagnosis, income, and insurance type — leaving too many patients without consistent access.

What the Bill Would Do

Introduced by Senators Wicker and Klobuchar and Representatives McGovern and Rutherford, this bipartisan legislation would:

- Require Medicare, Medicaid, CHIP, and the FEHBP to cover medically necessary foods and formulas as treatment.
- End the arbitrary requirement that patients use feeding tubes to qualify for coverage.
- Apply only to patients for whom physician-prescribed nutrition is their actual medical treatment.

Take Action Now

Congress needs to hear from you. Contact your representatives and urge them to cosponsor this legislation — then visit the action page to send a message directly:

nutritionequity.org/takeaction

MSUD Moves 2026: Walk, Bike, Run Together We Can Make a Difference!

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

MSUD FAMILY SUPPORT GROUP
MOVES
WALK • JOG • RUN

MSUD Moves is back—and we hope you'll join us this year! MSUD Moves is a community-driven awareness and fundraising campaign that brings individuals and families together to walk, bike, or run for MSUD—in whatever way works best for you. It's not a race, it's a movement. Every step, every share, and every donation helps raise visibility for MSUD and fuels the programs and progress our families rely on.

The best part? MSUD Moves is fun, flexible, and easy to do! You can host something as simple as a neighborhood walk, a park meetup, a school-track stroll, a family bike ride, or even a "dance-it-out" movement day with friends. Whether your group is big or small, you're creating something powerful: community connection, awareness in your hometown, and momentum for our mission.

Planning to participate on your own? Here are a few easy, meaningful ways to have a solo MSUD Moves event:

- One mile walk challenge: Pick a day, choose a route, snap a photo, and share why you're moving.
- "Lunch break laps": Walk a local loop for 20–30 minutes and invite friends to support your goal.
- Weekend bike ride for awareness: Ride your favorite trail and ask supporters to sponsor your miles.
- Sunrise or sunset walk: Enjoy a quiet moment of movement and reflection—then share a photo afterward to help spread awareness.

Funds raised through MSUD Moves will support the work that matters most to our community, including research initiatives, the MSUD patient registry, and programs that improve quality of life for individuals and families living with

MSUD. Last year, over $55,000 was raised to support the symposium taking place in June and research to further the mission of the MSUD Family Support Group.

If you're unsure where to start, don't worry—we've built simple tools to guide you through it. The MSUD Moves Fundraising Toolkit provides step-by-step support, with planning checklists, promotion ideas, sample social posts, email templates, a press release template, and more. Please email Denise at execdirector@msud-support.org to get a copy of the toolkit. You are never doing this alone—when one of us succeeds, we all do.

Will you move with us this year? You can participate throughout the year or choose to unite with MSUD families in late fall to enhance our joint impact in October. Join MSUD Moves, invite your family and friends, and help show the world what our community can do—together.

Questions? Contact Denise L. Kolivoski, MBA, Executive Director, MSUD Family Support Group at execdirector@msud-support.org for more information.

2026 MSUD Symposium

Don't miss the 2026 MSUD Symposium which will be held June 25th-27th at the Embassy Suites by Hilton Cleveland – Rockside in Independence, OH (just south of Cleveland). The symposium has something for everyone including new, seasoned and transplant families!

Check out the MSUD website at msud-support.org for information about how to register, make a hotel reservation, and view the agenda.

If you prefer to register by paper rather than online, please see the mailing that was sent to families in our database from the US, Canada, and Mexico early in 2026. It included information about the symposium as well as the printed registration form.

If you need more information or assistance with registering, please reach out to Sandy Bulcher at 740-972-5619 or sandybulcher@gmail.com

See you in Cleveland in June!

Sandy Bulcher, MSUD Symposium Coordinator

Rare Disease Day 2026:
Our Community Showed Their Stripes for MSUD

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

Rare Disease Day gave our community an opportunity to pause and recognize the strength, resilience, and everyday commitment of individuals and families living with MSUD. This year, supporters marked the day in a simple but powerful way: they wore stripes, snapped a photo, and helped make MSUD visible. Each image represented more than a moment—it reminded all of us that MSUD is not rare to the families living it every day.

We were honored to feature our 2026 "Show Your Stripes" participants using the hashtag #ShowYourStripesForMSUD, including the Covington family, who celebrated one year of health since learning about Crew Covington's diagnosis; a photo of Sandy Bulcher and Dave Bulcher, MSUD Board members and longtime champions for our community; and Ziva, showing her smiling face from Connecticut that reflected the care, teamwork, and pride that families and supporters brought to this journey each day.

While Rare Disease Day was a moment to be seen, it was also a reminder that visibility must lead to action. With that in mind, our organization continued the momentum in March by hosting a free Advocacy Webinar Series that was open to all, designed to help families and supporters build practical advocacy skills and turn lived experience into impact.

The two live, virtual sessions focused on both the fundamentals and the personal. Workshop 1, "What Advocacy Is and How to Get Involved," covered advocacy basics and realistic ways to engage. Workshop 2 explored storytelling—how to shape a clear message, share it with confidence, and connect effectively with policymakers and partners.

Jordann Coleman, MSUD Board Member and Advocacy Chair, reinforced why taking part matters and why advocacy is strongest when it reflects the voices of those living with MSUD every day. As Jordann shared, "Advocacy isn't just something we watch from the sidelines—it's something we do together. When our community speaks up, we help shape the policies and protections our families need, now and in the future."

This collaborative series was presented with HCU Network America and the Propionic Acidemia Foundation, in collaboration with EveryLife Foundation for Rare Diseases. We are grateful to everyone who attended, shared the sessions with others, and helped strengthen advocacy across rare disease communities. Rare Disease Day may happen once a year, but our commitment to visibility, community, and meaningful progress continues every day.

MSUD Diagnosis without Carrier Parents?

By: William Burns, MD, Geneticist at Nationwide Children's Hospital

Gerald is a unique patient case of MSUD, because he has no genetic mutation for MSUD, nor do his parents. At Nationwide Children's Hospital, the first time Gerald and his family saw the metabolic team was in the hospital for an abnormal amino acid profile at age 15 months, when Gerald had originally come to the hospital because he was not gaining weight as expected, was vomiting repeatedly, and showed signs of low muscle tone. During his admission, he had seizure concerns and got further work-up with metabolic labs. This led to the identification of allo-isoleucine and branched chain elevations in his blood, which were confirmed with multiple repeat levels and improved with dietary restriction of BCAA. Genetic testing was ordered and found to be negative after genome sequencing and microarray.

While he was still in the hospital and closely monitored, Gerald briefly returned to a normal diet without thiamine supplementation because his genetic testing was negative and his levels had improved. With this reduction in levels, his care team slowly introduced more protein into his diet to determine his tolerance. While inpatient, Gerald experienced a short time with a normal diet and no thiamine supplementation while in a safe environment, given his negative genome and levels. After about 48 hours, his levels increased again. Thiamine was restarted, and elevations resolved. With the lack of molecular findings but continued biochemical abnormalities, Gerald was diagnosed with thiamine-responsive maple syrup urine disease.

Gerald's diagnosis of MSUD has been strictly based on his biochemical results, which is rare, as it is estimated that less than 1% of patients with biochemical findings consistent with MSUD present with negative molecular testing. While research is ongoing to find the cause, whether it be a new gene or an undetectable molecular alteration, further work is needed. As with many other rare and ultrarare diseases, every single step is part of a complex medical odyssey.

Beyond his concerns for MSUD, Gerald has other health concerns, including pseudo-obstruction, a condition in which the intestines appear to be blocked, but without an actual physical obstruction. Throughout his treatment journey and multiple evaluations, Gerald has been seen by multiple genetics providers in other locations. Thus far, the consensus has remained molecular negative thiamine-responsive MSUD.

For now, Gerald remains on treatment for his MSUD, and his journey continues. His family has done a fantastic job with his care, and thiamine has played an important role in preventing further metabolic crises.

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Add 1-2 Tbsp Orange flavored syrup*

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CHOCOLATE
Add 1-2 tbsp (15-30 mL) chocolate flavored syrup*

STRAWBERRY
Add 1-2 tbsp (15-30 mL) strawberry flavored syrup*

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*Adding flavorings to COMPLEX MSD® Essential may change the protein content. Be sure to read labels or consult your healthcare professional and account for any additional protein.

COMPLEX MSD® Essential is a specialized formula from Nutricia North America for the dietary management of proven Maple Syrup Urine Disease (MSUD) and must be used under medical supervision. Not suitable as a sole source of nutrition. Suitable for individuals over 1 year of age.

NUTRICIA

Living Life with Classic MSUD from Childhood to Young Adult

By: Megan Greer

I want to share my story and my life now as a woman at age 24 and who has been married for about a year now. I was diagnosed with classic MSUD when I was 2 weeks old. Growing up my parents were my biggest advocates as well as my sisters (neither of them have MSUD). They supported me in everything I did then and now to this day.

When I was going through school I always tried to keep up with my sisters, every sport she did I wanted to do too. So I played soccer when I was in elementary school, then I signed up for soccer when I was in middle school, and was in the school band. During all of this activity I was able to maintain my levels with adequate hydration, calories, and protein throughout the day. I would drink some of my MSUD formula before going to school, but throughout the day at school I would eat my low protein diet, and would drink a high calorie either juice or gatorade. For dinner I would eat a low protein dinner, and then right before bed I would drink the remainder of the MSUD formula from the day. On days I was extra active I would drink extra MSUD formula shortly after finishing my activity.

I sometimes had a hard time with childhood illnesses like strep throat and ending up in the hospital. When I got to high school, I was in the high school marching band and played the keyboard and in the winter I did winter guard. I called that my "3 hour workout" twice a week every week for about 3-4 months every year for 4 years. My parents made sure I took in enough calories and drank my formula so that my levels stayed normal.

Now I am in that stage of life where I have gotten married and started a new life with my husband. I am learning to navigate life and keeping all my levels in check on my own with my husband, but of course I still have my family close by to help me when my levels get out of control, and I get super sick. That has only happened twice recently, first when I broke my ankle and had surgery. Due to this stress it caused my levels to elevate which we got under control in a couple of days never having to be hospitalized. The second time was when I had a stomach flu which took about a week to get my levels back to being normal.

I wouldn't change a thing. I appreciate everything I had growing up. I love that I was fortunate enough to get to try all the sports and try all sorts of new things throughout my childhood. I would recommend that other kids be as active as they can be, have fun and be a kid for as long as possible. I was fortunate enough that I was never limited on activities throughout my childhood, my parents helped me maintain healthy levels by monitoring my food intake even with all of my sports activities, as long as I consumed enough fluids and calories to make sure my levels didn't elevate. Everyone's tolerance is different and levels would need to be watched closely to keep them within adequate safe ranges.

MSUD RESEARCH UPDATE

By: Karen Dolins, MSUD FSG Research Lead

Everyone is eager for scientific breakthroughs that could truly improve our lives. As the Research Lead for our support group, my role is to promote research focused on better MSUD treatments and keep up with developments in the field. We have good reason to feel optimistic.

Petunia, the MSUD cow treated with gene therapy, continues to thrive. The team managing this project plans to continue their work by implanting new MSUD embryos so that gene therapy can be tested in more animals.

Researchers at Imagine Institute in Paris, France, in collaboration with Genethon (Evry, France), are developing novel gene therapy approaches for MSUD using laboratory models. They hypothesize that targeting skeletal muscle rather than the liver as in their initial proof-of-concept studies could overcome the limitations of liver-directed approaches and make adeno-associated virus (AAV) muscle-directed gene therapy a safer and more durable strategy. AAV is a leading tool used in gene therapy due to its ability to safely express genes in different body tissues.

Researchers at University College, London are working on two promising gene therapy treatments for MSUD. The first method uses an optimized gene delivery technology to treat the liver, with the aim of permanently restoring functional metabolism in liver cells for the rest of the patient's life. The second approach focuses on gene delivery to the brain, to investigate whether restoration of metabolism throughout the brain can alleviate neurological symptoms of MSUD better than treating the liver alone.

Syntis Bio has received NIH SBIR Fast-Track funding (R44DK143766) to advance development of a once-daily oral enzyme therapy for maple syrup urine disease (MSUD).

The program focuses on developing a gut-restricted enzyme that actively breaks down excess leucine in the digestive tract, helping lower leucine levels throughout the body. The therapy uses Syntis Bio's SYNT oral delivery technology to help keep the enzyme active in the gut for extended periods. This MSUD-focused effort builds on Syntis Bio's broader NIH-supported work developing oral enzyme therapies for rare metabolic disorders.

Home Leucine Monitor
We continue to work closely with biotechnology companies that are exploring the development of a device which would allow blood leucine levels to be assessed in the home. The goal is for a device which would provide results in less than 1 hour using a droplet of blood.

We're excited to announce that we will be holding our 3rd MSUD Science Summit in June. The meeting will bring together leading scientists, clinicians, and researchers who will participate in a day-long discussion of current work, ongoing challenges, and how best to move forward with MSUD research to improve care. Several of the participants will also be speaking at our MSUD Symposium. We will summarize their presentations in the Fall issue of this newsletter.

In the Professional Journals:
Gold, J. I., Grabill, M., Dolins, K., Basile, M., Hose, B. Z., & Ganetzky, R. (2025). Navigating Adulthood with Maple Syrup Urine Disease: Patient and Caregiver Perspectives on Healthcare Transition and Independent Living. Using a qualitative case study approach, the researchers assessed the lived experiences of adults by conducting extensive interviews with MSUD adults or their caregivers. Analysis of the interviews found an absence of standardized transition from pediatric care. A reduced emphasis on dietary management, a difference in symptoms of elevated leucine levels, and the need to address mental health and adult-onset conditions were also reported. Adult providers tend to lack knowledge of MSUD, providing a challenge to adult care. There is a need to provide appropriate support to achieve independent living.

"Metformin therapy to facilitate weight loss in adults with classic maple syrup urine disease" describes a study conducted by Rodrigues and colleagues at the Clinic for Special Children to be published in the journal Molecular Genetics and Metabolism (2026 Jan 7;147(3):109730. doi: 10.1016/j.ymgme.2026.109730). Eight adults with Classic MSUD were asked to reduce their caloric intake while taking metformin, a medication used to control blood sugar in people with pre-diabetes or diabetes, and which can aid in weight loss. After 1 year on the medication, participants lost a modest amount of weight while blood leucine remained stable. The researchers concluded that adults with MSUD can safely use this medication to aid weight loss.

Improving Diagnosis of Non-Classical Forms of MSUD
It is fortunate that maple syrup urine disease (MSUD) is included in newborn screening (NBS) in all US states. This enables physicians to rapidly diagnose infants born with the classic form of MSUD. However, NBS may miss non-classical presentations of the disease, including Intermittent and Intermediate MSUD, as blood leucine levels may not be elevated initially and the newborn may appear healthy.

Dr. Garrett and colleagues at Boston Children's Hospital describe their work using liquid chromatography-high-resolution mass spectrometry (LC-HRMS) to help identify MSUD in a 3-month-old baby who was previously healthy but later presented with an altered mental status, difficulty breathing, and seizures. LC-HRMS provided a comprehensive metabolic profile, enabling the analysis of a large number of metabolites (small molecules involved in the body's metabolism) from a single blood sample. Further analyses over the course of treatment showed changes in alpha-ketoacids, toxic compounds that build up in MSUD, along with changes in the branched-chain amino acids leucine, isoleucine, and valine. The researchers concluded that LC-HRMS may be a valuable tool for identifying metabolic diseases with non-classical presentations, such as this case of MSUD, and for monitoring the response to treatment in Classic and non-Classic types.

MSUD FSG Research Lead Dr. Karen Dolins communicated with Dr. Garrett, who advised her that currently LC-HRMS is mostly used as a research tool, but labs in Europe are starting to use it in routine screens. Boston Children's Hospital hopes to be able to implement this technology in the near future.

Garrett, R., Pickett, S., Peters, M. J., Belhassan, K., Ptolemy, A. S., & Peake, R. W. (2025). Application of high-resolution mass spectrometry profiling towards the diagnosis and acute management of maple syrup urine disease. Molecular Genetics and Metabolism Reports, 45, 101250.

MSUD Female Study

MSUD Female Study
Survey Conducted by: Dr. Jessica Gold, Northwell Health
Questions or to take survey by phone: jgold4@northwell.edu, 516-234-0763.
Survey: https://redcap.link/MSUDSRH

We are conducting a research study about how young adults with inherited metabolic disorders (IMD) make decisions about reproductive healthcare from puberty to contraception, pregnancy, and menopause. This topic is very important because many young adults with MSUD are now reaching adulthood, entering romantic relationships, and starting to think about starting families. This survey should be completed by women aged 18 years and older or caregivers to women aged 18 years and older with MSUD. It should take 15-20 minutes to complete.

You may have seen this survey posted on our social media or received an e-blast about it.

Vitaflo™ MSUD express™ plus and MSUD cooler® products offer isoleucine, leucine, and valine-free formulas with essential and non-essential amino acids, carbohydrate, vitamins and minerals.

Our range of products for the dietary management of Maple Syrup Urine Disease (MSUD) provides options to fit the demands of a modern lifestyle.

Unflavored provides flexibility to prepare a variety of delicious drink options.

MSUD express plus15
MSUD express plus20

- ✓ Suitable from 3 years of age
- ✓ Pre-measured powdered formulas containing 15 g or 20 g protein equivalent per packet
- ✓ Mix with 90 mL (3 fl oz) of cold water or other permitted beverages

Ready-to-drink options on the go for school, work, or travel.

MSUD cooler15

- ✓ Suitable from 3 years of age
- ✓ Ready-to-drink formulas containing 15 g protein equivalent per 130 ml (4.39 fl oz) pouch
- ✓ Available in Red and Orange flavors

Single Dose Amino Acid options for MSUD providing:

- ✓ Accuracy
- ✓ Safety
- ✓ Optimal quality
- ✓ Convenience
- ✓ Pre-measured packets containing:
  - – 50 mg of isoleucine or valine
  - – 1000 mg of isoleucine or valine

For more information about our range of products and to request a sample, visit **www.vitaflousa.com**

FOR USE UNDER MEDICAL SUPERVISION

Be sure to check with your healthcare professional before making changes to your diet.
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Vitaflo
Enhancing Lives Together
A Nestlé Health Science Company
Certified B Corporation

HEARTS ENTERAL, LLC - DME

11 Rande Dr. Wayne, NJ 07470
web: www.heartsenteral.com
P. 877-659-5540. F. 973-387-1223

LOOKING FOR A PLACE TO ORDER YOUR MSUD FORMULA AND MEDICAL FOODS? THERE IS HELP FOR MSUD PATIENTS!

We Supply Patients

- Access to a full range of MSUD formulas
- Low-protein foods from Ajinomoto Cambrooke and PKU Perspectives
- Peace of mind that patients and families won't receive a bill as long as their insurance is paying a reasonable amount, changing the focus to caring for the patient

Why Hearts Enteral – DME Was Created

Hearts Enteral – DME was established to address significant gaps in the coverage and supply of medical foods and formulas for patients with rare metabolic disorders.

We Provide

- **Direct Support to Patients**:
    - **Assist families**: Our team works to obtain 100% coverage of medical formulas/foods through health insurance, in most cases even if only part of this coverage is met, families do not receive a bill. We work with all private insurance companies and some Medicaid companies. For more details on insurance please visit https://heartsenteral.com/health-insurance-plans-1
    - **Service Model**: Provide medical formulas/foods directly to patients' homes, ensuring uninterrupted access.
    - **Certified Medical Coders and Billers**: Our team members are certified professionals in medical coding and billing; they are proficient in navigating the complexities of medical terminology and insurance reimbursement processes This is vital in insurance coverage for medical foods and enteral nutrition. Our background in medical foods and formula allows us to advocate effectively for patient coverage and manage the intricate details of billing and reimbursement.
    - **Providing Comprehensive Support**: We assist patients in understanding their insurance benefits, finding suitable health plans, and ensuring their medical nutrition needs are fully covered.
    - **Ensuring No Surprise Bills**: We adhere to the Federal No Surprise Billing Act, ensuring transparency and protecting patients from unexpected charges.

Please note a prescription from a medical licensed doctor is required.

Notice: We are processing a Patent and trademark with the USPTO - U.S. Patent and Trademark Office

ANPAD's Camp Knot-A-Phe 2026: Survive the Jungle

CAMP KNOT-A-PHE

Survive the Jungle

ANPAD - Arizona Network for Pku and Allied Disorders and Phoenix Children's Hospital metabolic team presents Camp Knot a Phe

Who?

All metabolic patients and their families who have been prescribed a protein restricted diet with medical formula. PKU patients on treatment with Palynziq are welcome!

Where?

Prescott Pines 855 E School House Gulch Rd
Prescott, AZ 86303

Lodging

All dorms areas have bunk beds. Each family has their own private rooms. Groups of rooms share a common living space and bathroom.

Our Activities

- ✓ Zip line
- ✓ Archery
- ✓ Swiming
- ✓ Horseback riding
- ✓ Axe throwing
- ✓ Arts and crafts
- ✓ Activities for under 5
- ✓ Dance party
- ✓ Hikes
- ✓ Discovery group
- ✓ Workshops
- ✓ Meeting new friends

Cost?

Camp Knot a Phe, the registration fee for camp is per family. This include meals, logging and activities.

More info?

email- info@anpadnews.org
or jami@anpaddnews.org

Meals

All low protein meals and snacks will be provided by a qualified chef. Regular meals will also be provided.

Formula

All medical formulas and medicines must be supplied by the family. Dieticians and volunteers will be on-hand to assist with meal planning.

transportation?

Car pooling is encouraged.

Register on our website

anpadnews.org

Get ready to Survive the Jungle! ANPAD's beloved Camp Knot-A-Phe returns June 12–14, 2026, bringing together families from across Arizona for a weekend of connection, education, and unforgettable fun. Hosted by the Arizona Network for PKU and Allied Disorders (ANPAD) in collaboration with the Phoenix Children's Hospital metabolic team, this one-of-a-kind camp is designed specifically for individuals living with PKU and allied metabolic disorders, including MSUD, who follow a protein-restricted diet with medical formula, along with their immediate family members. Individuals on Palynziq treatment are also welcome.

Camp Knot-A-Phe is more than just a getaway... it's a place where kids, teens, young adults, and parents can truly feel understood. Located in Prescott AZ, families share experiences, build friendships, and learn from medical professionals who specialize in metabolic care. While campers enjoy themed activities, games, crafts, and outdoor adventures inspired by this year's jungle theme, parents gain valuable insight through informal discussions and access to trusted experts.

The "Survive the Jungle" theme symbolizes strength, resilience, and community—qualities that define families navigating life with metabolic disorders. From poolside fun and field games to creative stations and team challenges, every activity is thoughtfully planned to be inclusive and diet-appropriate. Most importantly, camp provides something priceless: the chance for patients to meet others just like them and realize they are not alone.

anpadnews.org About ANPADNews
(https://youtu.be/td2_nMCSKCI?si=FMIKjJMvwV42JC3h)

Save the Date!

ILLINOIS LOW PROTEIN FAMILY CAMP

Aug 7 - 9th, 2026

Great Oaks Retreat Center
1380 County Rd 900 N
Lacon, IL 61540

Fun activities & games for the whole family!

- Swimming
- Fishing
- Canoeing
- Paddle boating
- Archery + ziplining
- And more!

Join us back at the beautiful Great Oak Retreat Center for a weekend away with other PKU and IEM families. Memories will be made, meals will be shared and friendships will be fostered.

pku Organization of Illinois

Information : danaebartke@gmail.com

Registration coming soon...
https://www.pkuil.org/newsevents.html

Relax, Connect, Belong, WE GET IT!

CAMP HUBER

- **Free for individuals with PKU and Allied Disorders**
- $150 for ages 3+

JUNE 5–8, 2026

712 Pughtown Road,
Spring City, PA 19475

443-694-6425

camphuber94@gmail.com

REGISTER TODAY

Introducing APF CARES — financial assistance to help everyone access our programs and activities.

Save the Date

CAMP MAGNIPHEQUE

Exciting Activities for All Ages.

Campfires - Fun - Nature - Hiking

Saturday Sept 5th-
Monday Sept 7th
(Long weekend)

Saskatoon,
Saskatchewan

MORE INFO:

www.canpku.org/events/CM_EN

HCU, MSUD PKU and UCDs

Toll Free: 1-877-226-7581
(1-877-CanPKU1)

International attendance welcome

APPLY TODAY FOR THE SUMMER OF A LIFETIME!

Hole in the Wall offers **FREE** 6-day residential Summer Camp in Ashford, Conn. for children with serious illnesses aged 7 – 18. Campers participate in traditional camp activities while receiving onsite medical care from doctors and nurses!

From the pool to the boathouse and the horse barn to the theater, Camp's magical setting provides the perfect backdrop for traditional camp programming **adapted so all campers can participate.**

Can't get enough Camp? Check out our Spring and Fall Family Camps for Camp-fun for the whole family! Eligible for diagnosed campers aged 5 - 15.

APPLY FOR SUMMER CAMP TODAY!

www.holeinthewallgang.org/apply

www.HoleintheWallGang.org | admissions@holeinthewallgang.org | (860) 429-3444

MSUD
FAMILY SUPPORT GROUP
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Columbus, Ohio 43235

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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 editor msudeditor@gmail.com or call/text Susan at 781-420-2676.