WWW.MSUD-SUPPORT.ORG

MSUD NEWSLETTER

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UNITED FOR LIFESAVING RESEARCH:

A COLLABORATIVE CALL TO DEFEND NIH FUNDING

The MSUD Family Support Group believes that research is the key to improving the daily quality of life for individuals and families living with MSUD. Recently, we championed support for research by drafting a letter advocating for sustained federal investment in rare disease research—a letter that has been endorsed by several allied organizations. This collaborative effort reflects our shared commitment to advancing treatments and improving outcomes for those affected by rare diseases.

We are dedicated to advocating for individuals and families impacted by rare diseases, we express our deep concern over the proposed funding cuts to the National Institutes of Health (NIH). The suggested policy change to cap indirect cost funding threatens to strip essential research institutions of the resources they need—resources that are vital for maintaining laboratory space, equipment, and staffing. Without these critical supports, groundbreaking research that provides hope to millions of Americans could be delayed or even halted.

Rare disease research already faces unique challenges due to the limited number of patients affected by each condition, making sustained federal funding an indispensable lifeline. The NIH has long been instrumental in driving discoveries and therapeutic advancements, particularly for conditions that receive little private investment. A reduction in funding would not only jeopardize current research projects but also undermine future innovations, with serious repercussions for the broader rare disease community.

Beyond the impact on research institutions, the proposed funding cuts carry significant economic consequences. Rare diseases impose a heavy financial burden on society, with per-patient costs far exceeding those of more common conditions. Reducing NIH funding risks exacerbating these challenges by stalling progress that not only saves lives but also sustains economic stability through job creation and continued innovation in medical research.

Your support sends a powerful message that research is essential for advancing treatments and improving the quality of life for those affected by rare diseases. We encourage you to send this letter to your representatives, underscoring our shared commitment to defending lifesaving research. Together, we can ensure that the momentum of scientific discovery continues and that our nation remains a leader in medical innovation.*

Inside This Issue:

The information contained herein does not neccessarily represent the opinions of the MSUD Board, Medical or Nutritional Advisors, or all of our members.

Before applying any of the information contained in this newsletter, you must consult a MSUD specialist.

Sections

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 _____

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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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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.

MSUD
FAMILY SUPPORT GROUP

We now have 209 participants from 17 countries!

Completing the MSUD Registry is one thing you can do TODAY to improve the lives of those with MSUD and their families.

Your participation in the registry will help with:

- Finding a cure
- Developing better treatments
- Improving quality of life
- Identifying common symptoms and impacts of the disease
- Sharing successful management experiences
- Aiding in the development of MSUD guidelines
- Supporting parents of newly diagnosed children
- Improving self-care and independent living

Register Now!

Please take a few minutes to register by going to our website: www.msud-support.org/patient-registry or by going directly to the registry: http://research.sanfordhealth.org/rare-disease-registry

Hard copies and Spanish language copies of the questionnaire can be obtained by cords@sanfordhealth.org or calling 877-658-9192.

Update your information annually

The MSUD Registry will be most effective if it is updated annually.
Email cords@sanfordcords.org or call CoRDS at 877-658-9192 to update your information.

Thank you for taking the time to improve our knowledge of life with MSUD!

Remember, all information is de-identified so responses cannot be linked to your name or other identifying information.

Want to learn more about our registry? https://msud-support.org/patient-registry/

Questions? Contact Karen Dolins at 914-391-2982 or karen.dolins@gmail.com

FROM THE PRESIDENT'S DESK

By Sandy Bulcher

On a cold winter day in Ohio, I sat down to write this article for the newsletter but didn't know what I wanted to share with you all. After some contemplation, I decided to share our MSUD journey. My hope is that it inspires you to connect with others within our community and become involved in the support group.

My husband, Dave, and I recognized the importance of support at the beginning of our MSUD journey when our son, Jordan, was diagnosed with MSUD in 1989. I attended my first MSUD symposium in Montreal, Canada, in 1990 when Jordan was just six months old. Unfortunately, Dave was unable to attend because he stayed home to be with Jordan, who was ill at the time. After that first symposium, I gained an even deeper understanding of the importance of fostering and maintaining relationships within the MSUD community. Our parents and close friends were helpful and encouraging, but they couldn't truly understand what daily life was like with MSUD.

During Jordan's early years, we decided to make attending the MSUD symposium a priority and incorporated them into our family vacations. Every two years, we packed the van and drove with our two boys (our older son, Tyler, does not have MSUD) to the symposium destination. The boys grew up playing with other children with MSUD and their siblings at the conferences. I have fond memories of those symposiums; the connections we made were invaluable, especially when Jordan was ill and hospitalized, and I needed an MSUD friend to talk to.

After attending the 1994 symposium in Missouri, Dave and I felt prepared to tackle hosting a symposium, which we did in 1996. It was a great deal of work but incredibly rewarding. Since then, I have been involved in coordinating the symposium in some capacity, and eventually, both Dave and I joined the MSUD Family Support Group Board, where we continue to serve in leadership roles.

You may not aspire to host a symposium, but a small gesture—such as hosting a fundraiser, financially supporting the group, or getting involved in the organization in another way—is essential for our organization to continue growing and thriving, and it will likely be rewarding for you. We also encourage our transplant families to become involved, as you are important to the success of the organization.

Incidentally, Jordan has managed daily life with MSUD quite well overall. He is married to Ashley, and they live and work in the Columbus, OH, area not far from us. We are fortunate that Tyler, his wife Tess, and our grandson Reid also live in the area. After 35 years, living with MSUD is a way of life for him, and thankfully, most days with MSUD are uneventful.

As always, please reach out to me at sandybulcher@gmail.com or 740-972-5619 anytime if you have questions, comments, or concerns. See you at the next symposium or sooner!*

FROM THE DIRECTOR'S DEN

By Denise Kolivoski

It takes all of us working together to make a difference. This spring, I'd like to introduce you to one of our incredible volunteer MSUD Ambassadors, as she plays an important role sharing her family's story and supporting advocacy for the MSUD community.

On February 21, 2025, the MSUD Family Support Group joined forces with the National Organization for Rare Diseases (NORD)'s Pennsylvania Rare Action Network (RAN) for a powerful Rare Disease Day virtual event. One of the highlights of the event was a moving keynote address by MSUD Ambassador, Ashley Bricker, a dedicated mother and advocate who shared her family's journey with her son Johnny's life with MSUD. Ashley's heartfelt presentation shed light on the unique challenges of living with MSUD and the resilience required to navigate life with a rare condition. "Sharing Johnny's story was an incredible experience," said Bricker. "Events like this are so important for raising awareness and educating others about MSUD. They help create a stronger, more connected community while inspiring others to take action and support rare disease advocacy."

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The Bricker Family

In addition to Ashley's keynote, the event featured discussions on advocacy efforts, personal stories from other rare disease families, and opportunities to engage with the rare disease community. Attendees were encouraged to wear stripes to show support and to share their own stories to amplify awareness. Recognizing the importance of community engagement, the MSUD Family Support Group deeply appreciates Ashley's dedication and the impact she continues to make. Her passion embodies the spirit of our MSUD Ambassadors—volunteers who help spread awareness, share updates, and support our advocacy initiatives.

For those inspired to contribute to raising awareness in their communities, we invite you to consider becoming an MSUD Ambassador. You can learn more and get involved by visiting https://msud-support.org/contact/. Together, we can create a brighter future for the rare disease community.*

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

FROM THE EDITOR'S LAPTOP

STRUGGLES WITH FORMULA SUPPLY DISTRIBUTORS

By Susan Needleman

In recent years, many of us have encountered challenges in receiving our prescribed formula due to manufacturing backorders. However, beyond these supply shortages, I have personally faced additional obstacles related to obtaining my formula. These challenges have stemmed from issues with insurance, durable medical equipment (DME) providers (who distribute the formula), and compounding pharmacies.

Navigating these challenges can be particularly difficult because each state in the U.S. has different laws regarding insurance coverage for medical formula. It is important to note that some states provide 100% coverage year-round, while others limit coverage based on age, only cover a portion of the cost, or require patients to meet deductibles before coverage kicks in and/or pay copayments and coinsurance fees. Unfortunately, while I live in a state where my formula should be fully covered, my private insurance comes from a self-funded employer, meaning it is not required to follow state laws. As a result, I am responsible for meeting a deductible and paying coinsurance fees until I reach my out-of-pocket maximum—an amount determined by my insurance company.

Adding to these challenges, one of the formulas that I was on at the time I was searching for a new DME was not widely available through providers who carried MSUD formulas. Many DMEs did not stock this one, possibly because it was a new product. It is also important to note that not all DMEs accept the same insurance plans, which further complicates the process. While searching for a provider that carried my formula and accepted both my primary and secondary insurance, I called between 20 and 30 DMEs, only to find that none met both criteria. The best option I found was a DME that carried all of my necessary products but only accepted my primary insurance. This meant my insurance would cover 90% of the cost, but only after I paid a deductible and a 10% coinsurance fee. Once I reached my out-of-pocket maximum, my insurance would then cover the full cost for the remainder of the year.

In addition to the MSUD powder in my formula, I also supplement with amino acids—specifically isoleucine, valine, and alanine. For over ten years, I had been receiving these supplements from the same compounding pharmacy without any issues. However, last year, everything changed. It is important to recognize that even a reliable supplier can suddenly become unreliable, as was the case with my compounding pharmacy. I first noticed that they had mixed up the labels on two of my amino acid supplements. Soon after, they became short-staffed,

Continued to page 6

stopped answering calls, and failed to return messages from both my doctor and me.

Suddenly, I found myself in a situation similar to my DME search—calling around to various compounding pharmacies to find one that could prepare my supplements, accept both of my insurances, and properly bill them. After many inquiries, I eventually found a compounding pharmacy that only accepted my primary insurance but could make all of my amino acid supplements—once again leaving me with out-of-pocket costs until my insurance coverage threshold was met. It is crucial to remember that not all compounding pharmacies and DMEs have experience with billing for metabolic conditions, so persistence and follow-up are necessary to ensure proper coverage.

With the costs of both my formula and amino acid supplements quickly adding up, I had to find a solution. While commercial supplements for isoleucine and valine were available through my DME, I would still have to pay a 10% coinsurance fee. Moreover, alanine supplements are not commercially available, meaning I had no alternative but to source them from a compounding pharmacy. Ultimately, I was able to get assistance for both my MSUD formula and compounded amino acids through patient assistance programs, which have covered nearly all of my out-of-pocket expenses.*

Advice for Navigating Durable Medical Es, Compounding Pharmacies, and Health Insurance

If you find yourself needing to switch to a new DME, compounding pharmacy, or product, here are some strategies that I found helpful:

1. Take the initiative to call around. While metabolic clinic offices and manufacturers can help, making calls yourself often yields faster results and more options. Some DMEs will only communicate with doctors until a patient is formally enrolled. If one provider refuses to speak with you, don't be discouraged—keep searching for another option.
2. Inquire about patient assistance programs. Many DMEs, compounding pharmacies, and manufacturers offer assistance programs, so it's worth checking to see if they have this option and if you qualify.
3. If your insurance denies coverage, take action. Ask the DME or compounding pharmacy to appeal the denial, and follow up with all who are involved during the process.
4. Double-check your insurance coverage. Call your insurance provider to confirm coverage details. If something seems incorrect, speak with multiple representatives to verify the information, as different agents may provide different answers.
5. Ask your doctor about alternative supplement options. Some amino acid supplements come in different forms, and one form might have better insurance coverage than another.
6. Follow up with your DMEs, Compounding Pharmacies, and your metabolic clinic. Your clinic must send paperwork to the DME or compounding pharmacy, so check with them to ensure everything has been submitted and to request updates. Also, follow up with the DME or compounding pharmacy to confirm the next steps and what documentation they still need.
7. Follow up with DMEs or compounding pharmacies later that are in the process of becoming in-network with more insurance providers or gaining access to more products.
8. Always check the labels. Once you receive your products, carefully read the label on each can, packet, or bottle to confirm that you received the correct item. Mistakes can happen, and consuming the wrong product could have serious consequences for you or your child.

By taking these steps, you can improve your chances of securing the formula and supplements you need while minimizing unexpected costs and delays. It is important to advocate for yourself, stay persistent, and utilize available resources. If you are facing similar challenges, know that you are not alone—there are solutions and support systems in place to help.*

The Vitaflo™ Promise

There's a lot to consider when choosing nutritional products. At **Vitaflo**, we promise to be your trusted choice. Click on the video link to learn more about how we're living up to this promise and supporting the Maple Syrup Urine Disease (MSUD) community.

Watch Video Here

For more information about MSUD products and to request a sample, visit **VitafloUSA.com** or call 1-888-848-2356.

For our collection of low protein recipes, check out the recipes section on our website **VitafloUSA.com/recipes**

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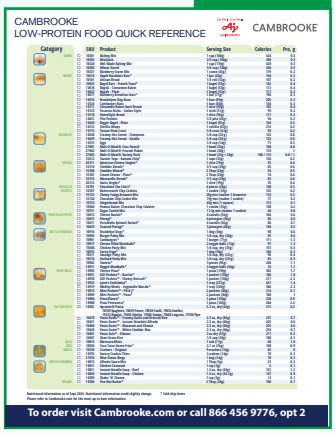

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PET PAGEANT 2025: A FUN & FURRY WAY TO SUPPORT THE MSUD COMMUNITY!

Back by popular demand, the MSUD Family Support Group Pet Pageant is returning in 2025! Last year's event exceeded expectations, raising an incredible $6,000 and bringing pet lovers together in support of families affected by MSUD. Thanks to the enthusiasm of our community and the generosity of our sponsors, we are ready for another round of friendly competition—all to benefit MSUD education, support, and advocacy.

How It Works

The Pet Pageant is a photo contest where participants can showcase their pets and encourage friends and family to vote. Each $1 vote serves as a donation to the MSUD Family Support Group, helping us continue our mission. The top two pets with the most votes will receive a special feature on custom wine bottles, and additional prizes will be awarded to other participants.

Prizes for 2025

This year's Pet Pageant is proudly sponsored by Nexus, Vannucci Vineyards, and the Coleman Family, providing incredible prizes for our winners:

1st Prize:

1. A custom wine label featuring the winning pet, with six bottles of wine (your choice of red or white) showcasing the label, generously donated by Vannucci Vineyards in Sonoma Valley, California. Winners can choose between red or white wine.
2. A $150 gift card to an online pet store to pamper your pet, donated by the Coleman Family.

2nd Prize:

1. The second-place pet will also be featured on a custom wine label and receive three bottles of wine (your choice of red or white) from Vannucci Vineyards in Sonoma Valley, California. Winners can choose between red or white wine. Learn more about the delicious wines from Vannucci Vineyards at https://vannucci-vineyards.com/.
2. A $100 gift card to an online pet store, donated by the Coleman Family.

3rd Prize:

1. One winner, selected through a random drawing (one entry per $100 raised), will receive a $75 gift card to an online pet store, donated by the Coleman Family.

How to Enter & Vote

1. Submit a photo of your pet and showcase their personality!
2. Encourage your friends and family to vote. Each $1 vote goes directly to supporting the MSUD Family Support Group.
3. Have the chance to win amazing prizes while raising awareness and celebrating our beloved pets!

Don't Miss Out!

Time is running out to participate—the contest ends in early May 2025! Every dollar raised through the Pet Pageant directly supports the MSUD Family Support Group's mission to provide education, support, and advocacy for individuals and families living with MSUD. You can take part in the contest here: https://www.gogophotocontest.com/petpageant Let's make the 2025 Pet Pageant another incredible success!*

CALL FOR

RESEARCH PROPOSALS

AND

APPLICATION GUIDELINES

MSUD
FAMILY SUPPORT GROUP
www.msud-support.org

The MSUD Family Support Group (MSUD-FSG) works to improve the lives of individuals with Maple Syrup Urine Disease (MSUD) through support, research, and advocacy. The MSUD Research Fund is a campaign of the MSUD-FSG to advance the science of MSUD by funding the most promising research that will lead to new therapeutic discoveries and a cure. We seek proposals that will help us meet the above strategic goals as well as close existing gaps in the knowledge and science of MSUD.

Our primary interests are:

- Identifying the impact of MSUD on the brain and developing therapies to improve neuro-psychological health.
- Developing therapies to improve the leucine tolerance of those with MSUD and reduce the toxic effects of elevated leucine and keto-acids.
- Developing a home monitor to assess blood leucine levels.
- Developing gene therapies to repair the defect in amino acid metabolism which causes MSUD.

We will, however, entertain any proposal which can be reasonably expected to advance treatments and improve the lives of those with MSUD.

Applicants are invited to submit project proposals meeting these goals using the guidelines presented in its *Request For Proposals*.

For questions related to MSUD research, contact MSUD Family Support Group Research Lead Karen Dolins at *karen.dolins@gmail.com* or *914-391-2982*.

NAVIGATING MSUD: A BRIEF GUIDE

By Nicholas Ah Mew, MD

The presentation titled 'Navigating MSUD: A Guide to Medical Management,' given at the MSUD Symposium on June 28, 2024, offered an overview of Maple Syrup Urine Disease (MSUD) for families new to the condition. Unfortunately, the video recording of the session did not come out. This article provides a brief summary of the talk. You can find more detailed information about MSUD at https://msud-support.org/about-msud/.

What is "Maple Syrup Urine Disease" and why does it have this name?
Maple Syrup Urine Disease (MSUD) is a rare genetic disorder that requires lifelong medical care. People with MSUD are missing an enzyme called Branched-Chain Keto-Acid Dehydrogenase or BCKDH. This enzyme acts like chemical scissors breaking down the 3 branched-chain amino acids: Leucine, isoleucine, and valine. These 3 amino acids are building blocks of protein that the body cannot make on its own, so we only get them from food. With normal food intake and not enough BCKDH, these 3 amino acids build up in the blood, which can lead to serious health problems. The disease is named for the sweet, burnt sugar odor found in the urine and earwax of people with MSUD.

Why is a high blood level of leucine dangerous?
Although blood levels of leucine, isoleucine and valine can all be high, only the high leucine is thought to cause health problems. High leucine can cause brain swelling (edema), which can cause brain damage. Symptoms of high leucine can include poor feeding, nausea, vomiting, irritability, weak muscle tone, unusual behaviors, seizures, and lethargy (excessive sleepiness).

How is MSUD inherited?
MSUD is passed down from parents in a special way called autosomal recessive inheritance. This means a child must get two changed (mutated) copies of a gene—one from each parent—to have the disorder. MSUD is caused by mutations in the genes BCKDHA, BCKDHB, DBT, and, less often, DLD.

How is MSUD diagnosed?
Early diagnosis of MSUD is very important to prevent serious health problems. In the U.S., all newborns are tested for this disease soon after birth through state-run newborn screening programs. Health care providers can also confirm the diagnosis with a blood test called a plasma amino acid profile, that will check for high levels of leucine, isoleucine, valine, and allo-isoleucine—a hallmark of the disease. Another type of test, called DNA sequencing, can find the exact gene changes in either BCKDHA, BCKDHB, DBT or DLD that cause MSUD. Some people with MSUD may try a special test called a Vitamin B1 (thiamine) challenge, where they take high doses of Vitamin B1 for several weeks to see if their leucine levels improve.

How is MSUD managed?
Every day, people with MSUD must get the right amount of leucine, isoleucine, and valine—enough to grow, but not enough to cause a harmful buildup. This balance is maintained through a strict diet, which often includes carefully controlled protein intake, a special formula without branched chain amino acids, extra valine and isoleucine supplements, and protein-free foods to provide necessary nutrients. Regular checkups are important. During these visits, health care providers check growth, development, health history, and blood levels of amino acids to adjust the diet and medicines as needed. Keeping blood branched chain amino acid levels within a safe range is important, especially leucine.

How can we prevent acute crises?
An acute crisis occurs when someone with MSUD experiences severe symptoms due to rapidly rising leucine levels, which can lead to brain swelling and increase the risk of brain damage. Preventing acute crises is an important part of managing MSUD. Common triggers like infections, not eating, or stress from surgery can change the body's metabolism and cause leucine levels to rise. To help prevent this, it's important to practice good hand washing and get regular vaccines, like the flu shot. It is also important for the metabolic care team to know if someone with MSUD is getting surgery (including dental procedures) and will need to not eat prior to the operation. The metabolic care team may suggest changes to treatment before, during and after the surgery.

In some cases, if someone with MSUD gets sick but is still awake and able to eat, they could follow a sick day plan, as prescribed by their metabolic care team. This plan will typically include cutting down on leucine intake, increasing the special formula without leucine, taking extra isoleucine and valine, and adding more calories and fluids. Medications can help with fever control and vomiting, and any infections should be treated quickly. But if someone with MSUD has trouble staying awake, can't stop vomiting,

Continued to page 12

can't eat, has high levels of ketones in their urine, or if the caregiver is unsure what to do, emergency medical care is needed. A sick day plan should only be used after discussion with the metabolic care team.

How do you treat acute crises?

The main treatment goal during acute crises is to decrease leucine levels as quickly as possible while managing other symptoms. This often requires the special formula that's missing leucine but includes other amino acids. Extra isoleucine and valine can actually be helpful in this situation. The formula and amino acids can be given by mouth or through a tube. Sometimes, if the person can't handle the formula, they might get a special IV solution that also doesn't have leucine. It is important to give them plenty of calories, either through the special formula or, if needed, through an IV. This helps reverse the body's breakdown of its own proteins for energy, which can release more leucine and make the MSUD crisis worse.

The fastest way to lower high leucine levels in the blood is hemodialysis, a medical treatment that removes toxins by filtering the blood through a machine outside the body. Dialysis can remove leucine quickly, but it needs special equipment that can take hours to set up, special IV access, and trained doctors, nurses and technicians. Because of the risks with dialysis and the long, complicated set-up, it is not always the best treatment option.

If brain swelling is suspected, which can be seen on a CT or MRI scan, other treatments like limiting fluids, giving special saline solutions or mannitol, and using medications to help remove extra fluid may be needed to reduce pressure in the brain.

What are long-term challenges in MSUD?

People with MSUD may face challenges beyond the daily treatment. They might have issues with development, like delays in learning, ADHD, anxiety, mood changes, and trouble with thinking and planning (executive functioning). These problems might require extra support, like therapies from early intervention programs or visits to specialists, such as brain doctors or mental health professionals. Some people also develop movement problems, like shaking, muscle stiffness, or difficulty with movement, which may also need special care. Despite these difficulties, women with MSUD can have successful pregnancies and healthy children if they carefully manage their health and leucine levels during pregnancy.

What's the role of liver transplantation in MSUD?

For some people, a liver transplant can be a long-term treatment for MSUD. The new liver will have a fully working BCKDH enzyme, which helps the body break down branched chain amino acids. The liver transplant allows most people to eat a regular diet and, in most people, it prevents acute crises. However, not everyone is a candidate for a transplant—it is only an option for patients who are healthy enough and have reached the right size and weight. While a liver transplant can greatly improve health and quality of life, it also comes with risks, like complications from the surgery and the need to take immune-suppressing medicine.*

TRAVELING WITH MSUD FORMULA – HELPFUL TIPS

Sandy Bulcher, President, MSUD Family Support Group

The cold weather in Ohio—and in many other parts of the country—has me thinking about warmer days and traveling. Traveling reminds me of the challenges we've experienced while journeying with our MSUD son, Jordan, and his formula. Typically, TSA removes his formula, examines it, questions him about it, and then returns it. Jordan carries a letter from his doctor stating that he will be transporting essential medical formula—whether in liquid or powder form—and emphasizing its importance for his health.

While I understand the necessity of TSA screening for questionable liquids or powders, the process can cause anxiety for those with MSUD and their families. There is always a concern that the formula might be confiscated, which could put the individual with MSUD at serious risk. I recently learned that programs exist to make the process smoother and less anxiety-inducing. TSA offers programs called TSA Cares and the Passenger Support Specialist (PSS).

According to TSA's website, TSA Cares is a dedicated helpline that provides information on screening procedures for individuals with disabilities, medical conditions, and other special circumstances. Assistance is also coordinated through the PSS program. PSSs are TSA officers who have received advanced training in assisting passengers with disabilities, medical conditions, or other special circumstances during checkpoint screening. A PSS can help from the moment you present your boarding pass and identification at the security checkpoint until you exit the area.

The level of service will vary based on the airport's resources. Some airports may call or email you ahead of time to coordinate a meeting point, while others may simply notify the checkpoint of your estimated arrival based on your departure time. You may also request to speak with a PSS or supervisor when you arrive at the checkpoint.

If you believe you would benefit from this service, please call TSA Cares at least 72 hours before your travel. They will collect your travel dates, flight information, and details about the assistance you need, then forward the information to the airport. If you are unable to provide 72-hour notice, you may request assistance from a PSS or supervisor at the checkpoint.

You may contact TSA Cares at 855-787-2227 between 8 a.m. and 11 p.m. ET Monday through Friday, or between 9 a.m. and 8 p.m. ET on weekends and holidays. You may also use the TSA Cares form available online at https://www.tsa.gov/contact-center/form/cares.

If you do not have a PSS with you at the checkpoint, TSA recommends separating your MSUD liquid or powder from your other belongings and notifying the TSA officer before screening begins. However, their website stresses that medically necessary items are not exempt from examination and will likely require additional screening. The container may be opened, but the TSA officer will not touch its contents. If you prefer that your container is not opened or x-rayed, you must inform the officer before screening begins. Choosing to have your medical formula hand-inspected will result in additional screening, including a pat-down.

Incidentally, I also learned that you are not required to provide documentation from your doctor to the TSA officer regarding traveling with medical formula. While verbal communication is encouraged, if you prefer a more discreet screening process, you can complete a TSA Notification Card. This card allows you to inform the TSA officer of any issues or concerns that might affect your screening. The Notification Card is available for download on the TSA Cares website. Please note that neither the Notification Card nor medical documentation exempts you from screening.

My hope is that these tips will simplify the process of traveling with MSUD medical formula. If you have any questions about traveling or any MSUD-related issues, feel free to reach out to me anytime at sandybulcher@gmail.com or 740-972-5619. Enjoy your upcoming travels!*

HAPPY 50TH BIRTHDAY JEFF FREDERICKS!

WRITTEN BY: ANNE FREDERICKS

Mother of Jeff, 50 Years Old, Classic MSUD, Pennsylvania

Fifty years ago, on February 17, 1975, Jeffrey Fredericks came into this world. He was a little early, but not a preemie. The first couple of weeks were normal, but then he started a very hurtful cry, and the arching of his back began. None of the doctors knew what was wrong because little was known about MSUD in 1975. After many visits to specialists away from our area, he was diagnosed with MSUD. It wasn't until we got to New York Medical Center that we were on the road to recovery with the correct diet. We were told that Jeff was so developmentally delayed that he would never walk or talk (what a joke!). We are very thankful for the care given by Dr. Holmes Morton. With the introduction of formula, the regulated food intake, and dedicated teachers, Jeff not only walks and talks, but has become an amazing adult with two part-time jobs. He is also a member of our township planning committee, on the board of the ARC, President of the local chapter of Aktion Club (a branch of Kiwanis), on a church committee, Honorary Mayor of our Development, and active in the PSU branch of Best Buddies.

Jeff has enjoyed being part of the MSUD Family Support Group, especially the Teen/Adult Zoom group. Our family has participated in numerous symposiums, and Jeff looks forward to being with friends who also drink formula and eat a limited diet as he does. It is truly a miracle that Jeff has survived to be 50. His hope is that a cure will be found soon.*

CALLING ALL OF THE MSUD COMMUNITY TO THE ALLIANCE OF PKU FAMILIES IN-PERSON EVENTS!

The Alliance of PKU Families is excited to welcome the MSUD community to Camp Huber and our PKU In the Park events for 2025! This is a fantastic opportunity for families and individuals with MSUD and similar conditions to connect, share experiences, and build lasting friendships in a supportive environment.

To join our email and mailing list to find out more about these events, contact us at:
https://tinyurl.com/APFmailinglist
443-470-5372

We look forward to seeing you at our events!*

CONOR MARTIN

BY JULIA MARTIN

Mother to Conor, 4 Years Old, Classic MSUD, Pennsylvania

Conor is one of four children of Daryl and Julia Martin and the only child in their family diagnosed with Classic Maple Syrup Urine Disease. Conor loves fishing and telling his latest fishing tales. He lives life with passion and makes friends with many he meets.

"Mr. and Mrs. Martin, we need you to come back to your son's room," the chaplain said kindly, but with urgency. "Your son has stopped breathing. The medical staff is working on him right now." My knees went weak. What was happening? Just hours ago, my life was perfectly normal. Then we received a concerning phone call from our family physician, informing us that our four-day-old son, Conor, had tested positive for Maple Syrup Urine Disease (MSUD) on his newborn screening. Praise God, the story doesn't end there. Although Conor had significant brain swelling by the time he was diagnosed with MSUD, he was spared a brain injury and recovered from his initial illness well. During those first months, my mind swirled with leucine levels, formula recipes, and a myriad of questions. Our life quickly shifted into a new normal—one where we constantly monitored amino acid levels and kept an ever-watchful eye for signs that Conor might be in a metabolic crisis. His first years were marked by numerous hospitalizations—about one every three months. Even though we tried to keep him healthy, he still caught the same illnesses any typical child might. The nurses at the hospital affectionately called him their "holiday baby" because it seemed like he spent every holiday with them.

At four months old, with guidance from Conor's medical team at The Clinic for Special Children, we made the decision to list him for a liver transplant. Our decision was based upon the understanding that a liver transplant could normalize his amino acid levels and protect him from the effects of a metabolic crisis. During the wait, Conor remained his usual mischievous self, indulging in a 5-pound block of cheese, sneaking ice cream, breaking an arm, and doing all the things little boys do. Some of these antics led to hospital stays due to MSUD complications, while others simply required a sick-day metabolic formula.

Two years after dual listing him at the University of Virginia and Children's Hospital of Pittsburgh, we received that monumental call. Conor received a very special gift that day in November. He was offered the left lobe of a cadaver liver. An adult gentleman, now known as Conor's "big brother," received the right lobe. The transplant recovery was challenging. Over the next seven years, Conor spent more days in the hospital than I care to count. He faced a critical bile blockage requiring reconstructive surgery, a bile duct stricture, and multiple episodes of organ rejection. Yet, through all of this, Conor learned that God knows him personally, cares about him, and has a plan for his future.

Today, Conor is celebrating his 11th birthday with cookies and cream cupcakes and has been hospital-free for two years. MSUD is still a term that we are very aware of in our household. That diagnosis will never go away, but the continual stress of knowing that Conor is at risk for neurological impairment is gone. No matter the medical management choice a family makes, we will always have a special bond with our MSUD family.*

A LETTER FROM LINDA LIVINGSTON TO THE MSUD FAMILY SUPPORT GROUP ON THE LOSS OF HER CHILD

Katy Livingston, Classic MSUD, born October 17th, 1970, in Kalamazoo, Michigan.

My daughter, Katy Livingston, passed away on August 21, 2024, in Birmingham, Alabama, after a lifetime of battling Classic MSUD. Katy's dad, Hank, passed just three months prior. We have consistently supported the group since its inception, though our participation in conferences was infrequent as we focused on life in Alabama. However, we remained engaged through the newsletter. Katy, a patient of Dr. Richard Allen, now deceased, of the University of Michigan, maintained a steadfast friendship with him throughout her childhood and adulthood. Every newsletter provided a valuable learning opportunity for the Livingston family, and I thank you from the bottom of my heart for your dedication and time in making this group meaningful to our MSUD community.

Katy's life taught us the value of community. Her life was meaningful to so many, and the people in her life meant the world to her. Katy was an incredible blessing to her dad and me.

Sincerely,
Linda Livingston
Katy's Mother

The following is an overview of Katy's wonderful life, written by MSUD Newsletter Editor, Susan Needleman, with the help of Katy's obituary and a tribute from one of her close friends.

Mary Kathryn "Katy" Livingston was born in Kalamazoo, Michigan, where she was diagnosed with Classic MSUD at just 11 days old. She was sent to Dr. Richard Allen at the University of Michigan, who had recently received funding to study MSUD. They became lifelong friends, even after Katy's family moved to Alabama when she was six years old.

Katy never let MSUD stop her from showing the world what she was made out of. She was an adored baby with a perpetual smile, drawing others in with her love of life. Despite the challenges that came with her disabilities, she faced them with determination. She wanted to do everything her siblings, cousins, and friends were doing and was always in the middle of their games and adventures. Katy also enjoyed participating in the Special Olympics, excelling in swimming and skiing.

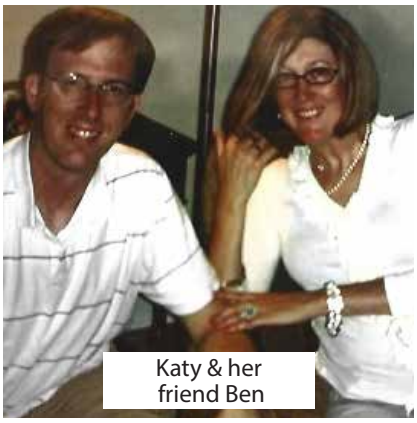
After graduating from high school, Katy moved to Birmingham and volunteered with AmeriCorps for two years. She was involved in activities at South Highland Presbyterian Church and worked at a deli while continuing to volunteer for various organizations. Katy lived independently in her condo for much of her adult life, with support from Triumph Services and others.

The Genetics team at UAB Hospital was a constant presence in Katy's life. Her generally happy nature made her beloved by the doctors, nurses, and specialists who worked with her over the decades—they became a second family to her. They also made sure she kept track of what she ate.

For Katy, family was everything. Before cell phones with saved contacts, she memorized the telephone numbers and birthdays of dozens of friends and family members. She faithfully stayed in touch with grandparents, siblings, aunts, uncles, cousins, friends, acquaintances, and the medical professionals who cared for her.
Katy had a life well lived.*


Katy and her father when she was born


Katy Graduating High School


Katy & her friend Ben


Katy at age 50

CARING FOR ADULTS WITH MSUD AND OTHER INBORN ERRORS OF METABOLISM

As treatment for inborn errors of metabolism (IEM), including MSUD, continues to improve and as adults with milder forms of these conditions are increasingly diagnosed, there is a growing need for practitioners with expertise in caring for adults with these diseases. In 2010, the Society for the Study of Inborn Errors of Metabolism (SSIEM) established the Adult Metabolic Physicians Working Group to enhance care for this patient population. In some countries, such as the United Kingdom, Canada, and the Netherlands, specialized departments dedicated to adult metabolic care have been developed. These clinics are staffed by providers trained in both adult medicine and metabolism, in contrast to the U.S., where many metabolic specialists have backgrounds in pediatrics.

In 2014, SSIEM's Adult Physicians Working Group published a report describing 6,182 patients seen across 24 centers worldwide. More recently, participating physicians from that study were asked to provide an update. Physicians from 15 adult centers responded, reporting care for 9,651 patients with 394 distinct diagnoses. Among these, 103 adults with MSUD were identified, three of whom were diagnosed as adults. This represents a 128% increase in the MSUD patient population since 2014, reflecting both improved life expectancy and increased diagnosis through newborn screening. Notably, in the discussion, the authors categorized MSUD as an inherited metabolic disease (IMD) that is not life-limiting.

Most adult metabolic clinics have experienced significant growth over the past decade. The IMD community has long assumed that the expansion of newborn screening in the early 2000s would lead to more IMD diagnoses, improved care, and increased life expectancy. This study confirms that assumption. The rise in adult patients is likely due to both enhanced patient care and longer life expectancy, as well as greater diagnosis rates through newborn screening. However, the true growth in adult patients may be even larger than reported, as 12 of the clinics that participated in the 2014 survey did not participate in this follow-up study, and only one new clinic was included.

As these adult patients transition from early adulthood into later life stages, their medical needs will differ significantly from those of the pediatric population. It is essential that healthcare providers develop the necessary expertise to support these patients throughout their lifespan.

Reference: Tchan M, Lehman A, van Dussen L, Langendonk JG, Janssen MCH, Langeveld M, Murphy E, Ryder B, Glamuzina E, Merkel M, Sechi A, Arnoux JB, Mochel F, Alkemade G, Maillot F, Kaphan E, Mazodier K, Thomas Q, Leguy-Seguin V, Marelli C. The Frequencies of Different Inborn Errors of Metabolism in Adult Metabolic Centres: 10 Years Later, Another Report From the SSIEM Adult Metabolic Physicians Group. J Inherit Metab Dis. 2025 Mar;48(2):e70005. doi: 10.1002/jimd.70005. PMID: 39912519.*

ACUTE METABOLIC DECOMPENSATION AFTER LIVER TRANSPLANT IN A PATIENT WITH MAPLE SYRUP URINE DISEASE

SUMMARIES FROM PROFESSIONAL JOURNALS

Summarized by Karen Dolins, EdD, RDN

Liver transplantation has become an effective alternative to dietary control in the treatment of Maple Syrup Urine Disease (MSUD). Instead of relying on metabolic formula and a rigid diet to regulate branched-chain amino acid (BCAA) levels in the blood, the new liver provides enough of the defective enzyme to process protein from natural foods. This typically prevents metabolic decompensation during illness. Some individuals feel they are "cured" of MSUD, although the transplanted liver provides only 9–13% of the defective enzyme, and BCAA levels generally remain above normal.

During illness, blood BCAA levels increase due to the breakdown of body proteins. This issue is further compounded when individuals are unable to consume adequate calories, which often occurs during gastrointestinal illness. If untreated, BCAA levels can continue to rise, leading to metabolic decompensation. While transplantation significantly reduces this risk, most post-transplant patients do not follow a sick-day diet.

This report describes the case of a 7-year-old boy with Classic MSUD who underwent liver transplantation at age 2. Before being admitted to the hospital, he had experienced vomiting and poor intake of food and fluids for three days. He presented with slurred speech and difficulty walking, and was diagnosed with encephalopathy (brain dysfunction) along with extremely high BCAA levels. His treatment included removal of dietary protein and administration of intravenous (IV) fluids containing glucose (sugar) and lipids. His leucine levels normalized within 22 hours.

Although metabolic decompensation after liver transplantation is rare, several cases have been reported. The authors suggest that families should be informed of this possibility and that amino acid levels should be checked during illness. They also recommend that IV fluids and glucose be administered promptly, particularly if there is concern for dehydration.

Reference: Tu SC, Khan M, Wolfe K, Kulkarni SS, Toolan E, Grange DK. Acute metabolic decompensation after liver transplant in a patient with Maple Syrup Urine Disease. JIMD Rep. 2024 Nov 25;66(1):e12460. doi: 10.1002/jmd2.12460. PMID: 39723122; PMCID: PMC11667767.*

TEACHING CLINICIANS ABOUT MSUD

Dr. Karen Reznik Dolins, EdD, RD, MSUD Family Support Group Research Lead and Board Member, recently served as an expert panelist for a Genetic ECHO session focused on MSUD.

The electronic Genetic Nutrition Academy (eGNA) was established in 2017 by Rani H. Singh, PhD, RD, at Emory University School of Medicine, Department of Human Genetics, to address gaps in metabolic specialty training among metabolic dietitians and other clinical providers who care for patients with inherited metabolic disorders (IMDs).*

MSUD AND THE BRAIN:

RESEARCH FROM THE LAB OF MSUD SCIENTIFIC ADVISORY BOARD MEMBER, DR. REBECCA AHRENS-NICKLAS

Investigators in the laboratories of Dr. Brian White and Dr. Rebecca Ahrens-Nicklas at The Children's Hospital of Philadelphia have a newly published study investigating neurological defects in a brain-specific mouse model of MSUD. These mice were bred to have normal amino acid levels in the blood, but lack the enzyme needed to metabolize branched-chain amino acids in the brain. Specifically, they used an innovative neuroimaging approach called widefield optical imaging. The technique can measure patterns of activity across the brain by detecting the changes in the state of oxygenation of blood. Unlike traditional MRI which produces static pictures of the brain, this allows investigators to study patterns of electrical activity in the brain. Mice that were missing the gene that causes MSUD in all brain cells or just astrocytes (one major type of brain cell), both showed abnormal patterns of activity. Specifically, there was increased coordination of activity between both sides of a key brain region, the somatosensory cortex. Increased coordination can sometimes be a proxy for seizure risk. Patterns of connectivity were further changed when MSUD mice were placed on a high protein diet. In summary, this work suggests that next generation imaging techniques that measure patterns of electrical activity in the brain may be useful in detecting MSUD severity and response to dietary or therapeutic interventions.

Lavery S, Adepoju TE, Fisher HB, Chan C, Kuhs A, Ahrens-Nicklas RC, White BR. Functional connectivity changes in mouse models of Maple Syrup Urine Disease. Cereb Cortex. 2025 Feb 5;35(2):bhaf040. doi: 10.1093/cercor/bhaf040. PMID: 40037414; PMCID: PMC11879283.*

THREE MSUD SCIENTIFIC ADVISORY BOARD MEMBERS CONTRIBUTE TO GENE THERAPY RESEARCH PAPER

If you've been reading our newsletter regularly, you know that the MSUD Family Support Group Research Fund contributed to the work of Drs. Jiaming Wang, Dan Wang, Heather Gray-Edwards, Kevin Strauss, and their colleagues to support their work investigating gene therapy in mouse and cow models of MSUD. The MSUD cow, Petunia, is now more than three years old and living out a normal life on a farm in rural Massachusetts.

The genetic mutation which causes MSUD can be found in any of three subunits (BCKDHA, BCKDHB, and DBT) which comprise the branched-chain alpha-ketoacid dehydrogenase (BCKDH) enzyme complex necessary for breaking down the branched-chain amino acids (BCAAs), leucine, isoleucine, and valine. Scientists from the UMass Chan Medical School, working in close collaboration with doctors at the Clinic for Special Children, designed and tested a recombinant adeno-associated virus (AAV) vector to deliver normal copies of the BCKDHA and BCKDHB genes to various tissues. This work is groundbreaking as it:

- Simultaneously replaces two of the three subunits that comprise the BCKDH complex and thus can be used to address the two most common forms of MSUD.
- Delivers the corresponding gene copies to multiple organs including the liver, muscle, heart, and brain.
- Utilizes both mouse and cow models of MSUD to demonstrate the safety and effectiveness of the therapy.

As in humans, most of the BCKDH enzyme activity in cows is found in muscle whereas mice express the large majority of BCKDH in liver. Using the cow model allowed researchers to track the uptake of the corrected gene in muscle tissue and assess its effects on chemical derangements caused by MSUD in the brain.

The researchers estimate that gene therapy increased the cow's weight-adjusted tolerance for leucine more than ten-fold. Blood BCAA levels were higher than normal after treatment (similar to what is observed after liver transplantation) but remained stable as the cow resumed a normal unrestricted diet high in protein. The mice and cows exhibited no liver or kidney toxicity from vector exposure and the cow has shown no behavioral or neurological deficits during more than three years of follow up. The UMass-CSC team is planning additional studies using a larger dose of the vector to determine if it enables even more decisive correction of blood and brain BCAAs.

Jiaming Wang et al. BCKDHA-BCKDHB digenic gene therapy restores metabolic homeostasis in two mouse models and a calf with classic Maple Syrup Urine Disease. Sci. Transl. Med.17,eads0539(2025).DOI:10.1126/scitranslmed.ads0539*

GRANT PROVIDED BY THE MSUD FAMILY SUPPORT GROUP SUPPORTING GENE THERAPY PROJECT IN MSUD CALVES

As described in previous newsletters, Drs. Kevin Strauss, Heather Gray-Edwards, Guangping Gao, Dan Wang, and Jiaming Wang have successfully treated an MSUD cow at the University of Massachusetts using gene therapy. The team requested a grant from the MSUD Family Support Group to help with the cost of maintaining the cows until they are able to secure an NIH grant to further their work. The request was favorably reviewed by our Scientific Advisory Board and a grant in the amount of $44,450 was approved by our Board of Directors. We are excited about the potential for this work.*

ASPIRE BIOSCIENCES AND UNIVERSITY COLLEGE LONDON

LONDON - NOVEMBER 28-29, 2024

Sandy Bulcher and Karen Dolins

MSUD Family Support Group President, Sandy Bulcher, and Research Lead, Karen Dolins, traveled to London to participate in Aspire Biosciences' "Collaborating for Change – Rare Metabolic and Liver Disease Cohort," held at the Royal Society of Medicine on November 28th (Thanksgiving Day in the United States). The event aimed to connect rare disease organizations with unmet needs to cutting-edge biotechnology companies interested in developing new therapies. In addition to our organization, approximately nine other advocacy groups and ten biotech companies were present. Each organization had the opportunity to briefly present information about their condition, highlight challenges, and discuss their most significant unmet needs. This was followed by short presentations from each biotech company outlining their platform.

Karen and Sandy were the only attendees from the U.S. The remaining participants came from the UK, Austria, Sweden, and Switzerland. They were joined by Dr. Robin Lachmann, a metabolic specialist from University College London (UCL), who volunteered to represent MSUD and provide a clinical perspective.

Representatives from three biotech companies expressed interest in working with our community. One company is developing an implantable liver device, another is attempting to insert the defective enzyme into red blood cells, and the third proposes using novel antibodies to neutralize the toxic byproducts of amino acid metabolism. All noted that their potential therapies are in very early stages of development. At this point, it is unclear whether these approaches will prove useful for MSUD. We have shared the information with our Scientific Advisory Board for their input before following up with these companies to determine if further collaboration is appropriate.

Dr. Lachmann's presence was invaluable. He asked insightful questions of the scientists and engaged in brainstorming sessions about the possible applications of their therapies for MSUD.

Before returning home, we met with Dr. John Counsell and his two doctoral students, who are conducting gene therapy research on intermittent MSUD mice. They shared slides of their current findings and provided a brief tour of their laboratory.

Both Sandy and Karen agreed that the event was highly worthwhile and are hopeful that meaningful progress will follow. At the very least, we have succeeded in raising awareness about MSUD among more biotech scientists.*

DR. REBECCA AHRENS-NICKLAS

Assistant Professor of Pediatrics, Children's Hospital of Philadelphia
Science Advisory Board Member, MSUD Family Support Group

The MSUD Family Support Group Scientific Advisory Board proudly introduces its newest member, Dr. Rebecca Ahrens-Nicklas.

Dr. Rebecca Ahrens-Nicklas is an Assistant Professor of Pediatrics at the Children's Hospital of Philadelphia and a valued member of the MSUD Family Support Group's Science Advisory Board. Her research focuses on understanding why patients with inherited biochemical disorders often develop severe neurological and cardiac symptoms that lack effective treatments.

By exploring the connection between biochemical imbalances and network excitability, Dr. Ahrens-Nicklas aims to identify novel therapeutic strategies that address the underlying causes of these conditions. Her work integrates fundamental biochemistry with clinical innovation, driving progress toward improved care for individuals affected by metabolic disorders.*

UNDERSTANDING THE MSUD BRAIN

By Kosar Khaksari, PhD

On July 28th and 29th, 2024, we had the opportunity to gather data from 21 individuals at the MSUD Family Support Group symposium. Our goal is to better understand brain activity in patients with MSUD, which will help improve their diagnosis and treatment. Among those who participated, 17 were MSUD patients and 4 were individuals without the condition who served as controls. The ages of all participants ranged from 4 months to 49 years. A big thank you to everyone who took part – we truly appreciate your involvement!

Since MSUD is a rare condition, gathering data from patients is one of the biggest challenges in our research. Being able to attend this conference and collect the necessary data was a crucial step, and it was a unique experience for our team. We are currently analyzing the results, but we need some additional information to interpret the data accurately.

If you participated in this study, you may have received an email from us requesting this information. We hope you will respond to this request so that we can complete our data analysis and let you know what we learned! If you are unsure of your genetic mutation, please ask your clinic if they have this information. It is also important for us to know the age at which symptoms first appeared.

We deeply appreciate your support and cooperation. Please don't hesitate to reach out to us with any questions.*

Kosar.khaksari@stjude.org
206 499 9389

SAVE THE DATE

CAMP CONNECT NOW OPEN TO MSUD!

September 5–7, 2025

Georgia PKU Connect is hosting the 14th Annual Family Georgia Connect Camp on September 5–7 (Friday–Sunday) at Camp Twin Lakes in Winder, Georgia, USA — centrally located in the Southeast and just 58 miles from the Atlanta Airport. Community members outside of Georgia are always welcome! In addition to parents and siblings, relatives such as grandparents, aunts, uncles, and cousins are also invited to attend! Camp activities include boating (kayaks, paddle boards, canoes, pedal boats), swimming in the pool, dodgeball, horseback riding, low-protein cooking, archery, shing, eld games, and other fun outdoor adventures.

Register at https://georgiapku.org/camp2025/
($50 per non volunteer camper)
Email camp@georgiapku.org or 678-612-8408 with any questions

COMMUNITY SUPPORT ALL YEAR LONG

BY SUSAN NEEDLEMAN, MSUD AMBASSADOR

As many of you know, the MSUD Family Support Group holds a symposium every other year, bringing our community together to support one another, to educate us on MSUD treatments and new advancements. With the help of the internet, this education and support continues year-round. We have a very active Facebook page and Facebook Group, monthly teen and adult virtual meet-ups, and, this past year, we also hosted two informative webinars.

The first one, Managing MSUD Diets in School, Daycare & at Home, was presented by Children's Hospital of Philadelphia (CHOP), dietitians Abby Opher Lauko, Taelyr Mello, and Shawn Carro on November 19, 2024. This presentation covered a wide range of dietary topics that many of us face. It guided viewers through every stage of a child's diet, starting at age one—when caregivers can begin discussing food choices with their children and teaching them to always ask if a food is safe before eating. The presenters emphasized the importance of fostering a positive relationship with the foods they can eat.

The webinar also addressed the challenges of the teenage years, when individuals with MSUD begin gaining independence with food and cooking. Throughout a child's schooling years, parents often struggle to educate schools about the MSUD diet, the necessity of formula, and the dangers of consuming high-protein foods. This session provided valuable tips on what to ask and inform schools about, as well as strategies for helping children navigate these situations. Additionally, it touched on how to cook for the entire family while accommodating a low-protein diet. As someone with MSUD who has personally experienced each of these life stages, already, I found the recommendations from CHOP insightful—some I wish I knew growing up. If you missed the webinar and would like to watch it, the recording is available here: https://youtu.be/_gBtHEs2SOM?si=wAgecd7vNUmV4iYv.

The second webinar focused on MSUD Health and Wellness and featured two segments—one from Ajinomoto Cambrooke and another from Zoia Pharma, a Pentec company. Ajinomoto Cambrooke provided a cooking demonstration using their Sugar Cookie Mix and shared helpful everyday cooking tips. Zoia Pharma, a Pentec company, showcased a cooking demonstration by Promin Metabolics, featuring Baked Macaroni and Cheese, Shepherd's Pie, and Chili Con Carne. All of the dishes looked delicious and made me hungry—I can't wait to make them myself! You can still watch this webinar here: https://youtu.be/FK4Vwx_2wws?si=nNFA-NallXyueH3T.

We'd like to thank the following sponsors for making these events possible:

- Zoia Pharma, a Pentec company
- Ajinomoto Cambrooke
- Children's Hospital of Philadelphia (CHOP)*

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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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