MSUD FAMILY SUPPORT GROUP

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MSUD NEWSLETTER

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STEP INTO ACTION: BE PART OF THE MSUD MOVES CAMPAIGN!

By: Danise L. Kolivoski, MBA, Executive Director

This fall, the MSUD Family Support Group is launching the MSUD Moves campaign, and we invite YOU to be part of it! Exercise is important for all of us, and in our rare disease community, every step has the power to raise awareness and bring us closer to better treatments and a cure.

Board President and MSUD family member Sandy Bulcher will be participating in Columbus, Ohio, and she encourages other families and individuals to join her:

"I'm excited to take part in MSUD Moves and walk with my family and friends in our community. It's a simple way to stay active, spread awareness, and support MSUD families. I hope you'll join me this September or October and be part of this important cause."

MSUD Moves is flexible and family-friendly — you can walk around your neighborhood, jog through the park, or gather your loved ones for a stroll in your community. Invite friends and relatives to support your efforts. Every mile and every donation brings us closer to advancing MSUD research and supporting the 21st MSUD Symposium. A special thank you to Nutricia for leading this national wellness initiative!

Contact Denise L. Kolivoski, MBA, Executive Director of the MSUD Family Support Group, to get started. She can send you a MSUD Moves Toolkit and help you along the way! Email execdirector@msud-support.org today!

SAVE THE DATE

MSUD **Symposium 2026** will be held June 25-27, 2026, at the Embassy Suites by Hilton Cleveland Rockside in Independence, Ohio (just south of Cleveland). Plan now to join us! Details about the symposium speakers and agenda will be available in early 2026.

DISBANDMENT OF THE FEDERAL NEWBORN SCREENING COMMITTEE

By: Jordann Coleman, Advocacy Chairperson

The disbanding of the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) by the Trump Administration in April 2025 has left a significant void in the U.S. newborn screening system. For over two decades, the ACHDNC played a crucial role in guiding newborn screening policy, ensuring consistency and scientific rigor across states. Before its establishment in 2003, screening varied widely, with some states testing for as few as four conditions and others for up to 50. The committee, in collaboration with the American College of Medical Genetics, developed the Recommended Uniform Screening Panel (RUSP), which became a national standard for newborn screening.

The ACHDNC's dissolution has halted progress on adding new conditions to the RUSP, potentially delaying diagnoses and treatments for thousands of newborns annually. This move has left states without federal guidance, exacerbating disparities between well-resourced and under-resourced states. The absence of a replacement plan has left ongoing nominations in limbo, with no federal mechanism to evaluate new evidence or guide policy.

Health advocates warn that this decision reflects a broader disinvestment in public health, threatening to undermine progress and innovation. Calls for reform, rather than disbandment, emphasize the need for a structured, evidence-based approach to newborn screening to prevent harm and ensure equitable healthcare for all infants.

The MSUD Family Support Group signed onto a letter authored by NORD with 271 other community organizations, including the American Academy of Pediatrics, American College of Medical Genetics and Genomics, American Society of Human Genetics, and Association of Public Health Laboratories, to the Secretary of Health & Human Services regarding the termination of the Committee and the elimination of other Federal Newborn Screening Infrastructure. We will continue to advocate for Newborn Screening at a Federal level. We encourage anyone who shares this concern to contact their representatives and explain why this leaves babies vulnerable.

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

FROM THE PRESIDENT'S DESK

By: Sandy Bulcher, Board President

Happy Fall! It's been a busy spring and summer for my husband Dave and I personally, as well as for the MSUD Family Support Group. In May, I retired after working more than 40 years as a registered nurse and in July, our sons and daughter in laws blessed us with two new grandbabies! Our adult son, Jordan with Classic MSUD and his wife Ashley, became the parents of Graham. It's extra special to see him as a father as there were times when we questioned what the future held for him. Our non-MSUD son, Tyler, his wife, Tess, and their three-year-old son Reid, welcomed their baby, Sylvie to their family.

In the spring, my family, friends and I participated in the spring MSUD Pet Pageant fundraising event and contributed to the $6,000 raised for MSUD. This is a fun event and one that I would encourage you to get involved in in the future.

Just after retiring in May, I participated as an exhibitor at an Abbott conference. It's important for our organization to attend events such as this to network with dieticians, other rare disease organizations, and biotech and pharmaceutical companies. We need to remind them of who we are, how we can help them and how they can help us.

In June, the MSUD Family Support Group board and our executive director, Denise Kolivoski, met in Norfolk, VA for our yearly face-to-face meeting. We discussed many topics, including growing our organization both physically and financially. It was a productive two days. During the meeting, we welcomed our new board member, Julia Martin, and celebrated Ivan Martin's long-term commitment to the board. He is transitioning off the board to emeritus status. Julia and her husband, Daryl, are the parents of Conor, who is 11, and was transplanted. Julia has connections to both the Mennonite and transplant communities and her input will be valuable as we look to better serve these communities. Ivan and his wife, Mary Kathryn, are the parents of Keith, who is 47 years old with MSUD. Ivan joined the board in 1998 and has served in several different positions over the years. Fortunately, our community will still benefit from his input, knowledge, and experience in his emeritus role.

This fall, I'll be working with a committee to determine the 2026 MSUD Symposium agenda and speakers. The event will be held in the Cleveland, Ohio area at the Embassy Suites Rockside in Independence, Ohio on June 25th-27th. Because of your input on the 2024 symposium surveys, we plan to provide more breakouts, social time, and transplant education for the 2026 symposium. Mark your calendars now and plan to join us!

I will also be participating in this fall's MSUD Moves campaign with the hopes of raising significant funds for the MSUD Symposium. My specific area of interest is fundraising to assist with expenses for those that would be unable to attend the Symposium otherwise. The symposiums are so valuable, because that is where long-term relationships are built and continue to grow. I hope that you will join me in participating in this campaign!

As always, if you have questions, concerns, or suggestions, please reach out to me via phone at 740-972-5619 or email at sandybulcher@gmail.com

See you in Cleveland next summer!

Jordan Bulcher (Classic MSUD) and his wife, Ashley, welcomed Graham Nicholas Bulcher on July 3, 2025.

FROM THE DIRECTOR'S DEN

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

As we approach the end of the year, I want to take a moment to reflect with gratitude on the incredible strength and resilience of our MSUD community. Every day, I see the ways our families support one another—sharing advice, celebrating milestones, and walking together through challenges that few outside our circle can truly understand.

This year, however, has brought us a difficult reality. Donations to the MSUD Family Support Group are down. While the need for education, advocacy, research, and family support continues to grow, the financial resources to sustain these efforts have not kept pace.

Families living with MSUD face medical costs, special dietary expenses, and daily challenges that stretch household budgets far beyond what most families ever have to consider. That makes it even harder to ask for your support. I also know this: without the commitment of our own community, the programs and progress we depend on cannot continue.

Your donation—whether $10, $50, or $100—makes a direct impact. It keeps our patient registry moving forward so researchers can better understand MSUD. It ensures families have a place to turn for trusted information and connection. It allows us to host webinars and symposiums that give hope and practical tools for daily life. And it shows the outside world—funders, partners, and policymakers—that our community believes in this mission and is worth investing in.

Even if your gift feels small, when added to others it becomes transformative. Imagine what we can achieve if every family gives just a little: together, we could strengthen research, provide vital resources, and create a future where those living with MSUD thrive.

I'm asking you today—before this year ends—to make a donation to the MSUD Family Support Group. Let's stand together, not only as families managing a rare disease, but as a community that believes in hope, progress, and each other.

Thank you for all you do to make this journey less lonely, more supported, and filled with possibilities.

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

FROM THE EDITOR'S LAPTOP

By: Susan Needleman

I like to use these editorials to connect something from my own life with something that might be happening in yours. For this issue, I want to talk about energy.

Many of us experience different energy levels throughout the day, and often this can be related to our amino acid levels—whether they are high, low, or normal. What I recently discovered is how important it is to share this kind of information with your nutritionist or dietitian.

For quite a while, I struggled with low energy for the first half of each day, even on days when my levels were normal. The change would happen during lunch, and I would feel like an energy light switch was suddenly put on in my body. I noticed that when this light switch was on, I could think sharper and had more energy, though it only recently occurred to me to mention it to my dietitian. When I finally did, we traced the change in my energy back to a new formula I had started. Working together, we were able to adjust the makeup of my formula, and now I have steady energy throughout the day—at least when my levels are within normal range. Of course, it's harder to manage when my levels are off, but being proactive has made a big difference.

Moral of the story: Share everything with your doctors and nutritionist or dietitian—even if it does not seem important. You might be surprised by how much they can help.

Sincerely,
Susan Needleman
MSUD Newsletter Editor

HONORING IVAN MARTIN'S SERVICE
TO THE MSUD FAMILY SUPPORT GROUP

By: Sandy Bulcher, MSUD Board President

Special thanks to Ivan Martin of New Holland, Pennsylvania, for serving on the MSUD Family Support Group Board since 1998. Ivan and his wife, Mary Kathryn, are the parents of Keith, age 47, who lives with MSUD, and Cheryl, who passed away in 1988 from complications of MSUD. They are also the parents of Marilyn, who does not have MSUD, and the grandparents of five.

After many years of dedicated service, Ivan has decided to step down from the Board of Directors. Thankfully, he has agreed to remain in an emeritus role. In this capacity, he can continue to be involved in the organization while we benefit from his many years of experience with MSUD, as well as his strong connection to our large Mennonite community.

Thank you, Ivan, for your commitment to the Board and to the MSUD Family Support Group!

JULIE MARTIN

New Board Member

I'm honored to join the board of the MSUD Family Support Group. After our now eleven-year-old son, Conor, was diagnosed with classic maple syrup urine disease (MSUD) through newborn screening, I became passionate about supporting families affected by this condition. Caring for him through his MSUD management and liver transplant journey has deepened my awareness of how important it is for rare diseases to receive greater recognition and support.

For the past seven years, I've worked as a Development Associate at the Clinic for Special Children in Pennsylvania, helping advance the Clinic's mission of providing compassionate, high-quality care for children and adults with rare genetic disorders.

At home, my husband Daryl and I are raising our four children. I enjoy serving at our church, spending summer days with my family at the beach, and exploring creative expressions through painting and photography.

I'm excited to bring both my personal and professional experience to the MSUD Family Support Group and support its mission of providing care, community, and hope to families living with MSUD.

To read more about my story please see the Spring 2025, issue of the MSUD Family Support Group Newsletter on page 15.

LIVING WITH MSUD IN THE OLDER YEARS
By: Sharlene Balinsky

50 Years Old, Classic MSUD
Montreal, Canada

I wanted to share with you what my life is like in my older years. As I get older, I've noticed that different issues come up. I have been struggling with my levels, which have been fluctuating for quite some time. This has been going on for several months, and when my levels are high, I sometimes feel dizzy. This past summer, I became extremely dizzy one day and couldn't walk without help—everything seemed foggy and blurry. I was very fortunate that my boyfriend, Andrew, was home at the time to help.

The dizziness continued off and on for about a month. At first, I thought it was due to high levels, which I had recently experienced, but it didn't make sense because I hadn't felt like this before. It turned out my levels had actually dropped from 700 to 350. My MSUD team said they didn't think it was related to MSUD. Even though dizziness can sometimes be a sign of high levels, my levels were normal. It is frustrating when MSUD symptoms overlap with

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something else, but that is the life of an MSUD patient.

I went to see my general practitioner (GP), who diagnosed me with Vertigo. While she was talking to me, I was looking at her, but my eyes were moving side to side, unable to focus. That told her right away that it was Vertigo. My GP had me try ear exercises to help realign the crystals (tiny calcium carbonates), in my inner ear that control balance. When she saw some improvement, she referred me to a physiotherapist who specializes in Vertigo. After just one session, my dizziness stopped.

Even before this, I had stomach issues for years. I saw several doctors, but no one could explain why I had so much pain and constantly felt uncomfortable and bloated. After years of dealing with this and going through many tests such as CT scans and X-rays, nothing showed up. While this was good news, I still wanted answers. I then tried some diet changes, based on my nutritionist's suggestions, but they didn't work. After a year, the doctors agreed to send me to a gastrointestinal (GI) specialist to rule out possible problems. In 2022, they performed both a colonoscopy and an endoscopy, together in one procedure.

During this procedure, my doctors were amazing in helping me prepare. They made sure I could tolerate the prep drink beforehand. I met with both my GP and my MSUD doctor to ensure they were aligned. My MSUD doctor, Dr. John Mitchell, at Montreal Urban Hospital for Children (MUHC), suggested checking my levels a few days before the procedure and again two days after. The day before, I ate normally, followed the clear liquid orders, and took extra formula to increase calories.

Dr. Mitchell knew the timing of my procedure and how long it would take. He emphasized the importance of scheduling the earliest appointment possible since I wouldn't be able to have formula for several hours afterward. I stayed in close contact with the clinic before and after the surgery, and my MSUD team was always available and checked in on me. The results were negative, and while the cause of my stomach pains is still unknown, thankfully the pain is not as intense as it once was.

As an adult with MSUD, I have also dealt with anxiety for years, sometimes leading to panic attacks. For a while, it calmed down but returned. In 2023, I began having hot flashes, and my GP prescribed a medication called "Lolo" to stop my periods and help with the hot flashes, thinking it could be premenopausal. I took it for about a year and a half. At first, it helped, but eventually it had the opposite effect. Since then, my doctors have been trying to find another way to manage my anxiety.

Only time will tell, but I find that as I get older, my levels fluctuate a lot more. My doctors and I suspect this could be related to perimenopause or menopause. Unfortunately, there isn't much medical knowledge on this, as the discovery of MSUD is still so recent that very few people with MSUD are in this age group. I am fortunate that my doctors are willing to learn, explore, and research to better understand our condition as we age.

It isn't easy, but they are trying, even with limited information. In a way, we are role models for the MSUD community. That's both encouraging and challenging, as we'd love to have all the answers—but for now, we continue to learn together.

PURSUING GOD'S GRACE

By: Libby Stone, MSUD Transplanted, Nebraska

Diagnosed at thirteen days old with MSUD, I was born into a world full of uncertainty, strict medical routines, and life-threatening challenges. In my new book, Pursuing God's Grace, I invite you to join me on my path of resilience, survival, and unshakable faith.

Through this journey, you are invited to walk alongside me as I navigate the turmoil brought on by my illness, leading up to two life-saving liver transplants, that took place in 2007 and 2024. Despite living with chronic pain most of my life and the countless setbacks I have experienced along the way, my spirit has remained unbreakable and my hope unextinguished.

I have been lost, scared, angry, and resilient. At my lowest points, I clung to hope and my faith. Through it all, God has always led me home.

This memoir is more than just a medical journey; it is a spiritual one. Having volunteered for eight years at a non-profit organization dedicated to advocating for organ and tissue donation and speaking at events and churches, I have committed my life to inspiring others with my message: never lose hope.

After being a year post-second transplant, life has changed in so many ways, yet somehow has remained the same. Since being home from the hospital, I am stronger than ever. I have a better connection with my loved ones. I have a new sense of hope and faith that I haven't experienced before. However, I am still having side effects from the surgery. My right lung collapsed a few days after getting home from my transplant. After having multiple surgeries in a month, with several drain tubes placed to help pull off the excess fluid, I am still dealing with chronic pain in my ribcage. My medical team has done everything they could to get me back to where my lung once was; however, it will never be 100% back to normal. I will never be pain-free because of the nerve damage that took place after removing the drain tubes. I am still hopeful that the longer I get

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post-transplant, the better I will feel. There is always hope, and I choose not to let my setbacks define me.

Set against the backdrop of my home in forested Bellevue, Nebraska, where I live with my supportive family and emotional support dog, Howie. My story is a reminder that our greatest trials and tribulations can become our greatest triumphs.

Pursuing God's Grace is a beacon for anyone battling chronic illness, questioning their faith, or searching for a deeper sense of purpose. It is a story of pain, yes- but more importantly, it's a story of praise.

Those living in the United States of America and Canada can purchase Pursuing God's Grace at www.iamelizabethstone.net or on Amazon https://www.amazon.com/Pursuing-Gods-Grace-Elizabeth-Stone-ebook/dp/B0F3WM1755.

DOCTOR SEARCH

By Nikolai Rudd

51 Years Old, Classic MSUD (transplanted), Massachusetts

Over the years, I have written many articles for this newsletter about the health challenges I have faced, especially since my liver transplant in 2006. This time I am writing about a healthcare problem facing all of us, whether you have MSUD or not. But that, of course, is a bigger challenge to the MSUD and MSUD Transplant community.

This past winter, I received a letter from my primary care's Office that said he was leaving by December 31st, and I would have to find a new doctor. However, the only doctor in the practice who had been accepting new patients, had already stopped.

So . . . I was effectively on my own and had to track down someone else in another practice. After calling Medicare and Blue Cross/ Blue Shield of Massachusetts I was given several possible leads. And was even given names of friends who were part of other practices. That being said, out of all the practices I called in western Massachusetts, I was able to find a few leads. Only four people were taking new patients. Because of so many doctors leaving the area or retiring, there is a severe shortage of doctors in my area. Which is very discouraging.

In the meantime, I tried to get some new prescriptions called into my pharmacy. But I was informed that "my doctor's office" received an email from the hospital administration that said they COULD NOT write any new prescriptions for me, since my primary doctor was no longer a part of their practice. My jaw dropped in disbelief! I was essentially hung out to dry. When I asked how I was supposed to get my prescriptions filled, they said I would have to drive to their Urgent Care facility. I had to get my immunosuppressant filled and my diabetes medication, as well as one of my pain medications. After waiting a couple hours, I was seen and got told by the doctor there that they couldn't fill my pain medications because it was a controlled substance. So, I ended up having to go to the ER at our hospital, just to get prescriptions for all my medications that were determined to be narcotics by the FDA. This "visit" took another three hours, but at least I was successful and hoped I didn't get sick from anyone, as I lay on a gurney in the hallway for the entire time.

When I talked to these offices, I was told that they were booking out appointments into October (nearly 10 months away). And they had already had 400 new patient applications for their one doctor. And that was before I could even get my hands on the application, fill it out, and drive there to turn it in. After not hearing back from the first practice, I luckily found two who were taking applications for new patients about an hour away.

Because of the doctor shortage in the mountains of western Massachusetts, the openings were with a Nurse Practitioner. To get around the shortage, the state started to allow Nurses and Physician Assistants to become Primary Physicians to patients. It was late February by now, and I jumped at the chance to schedule an appointment when I found out the first available appointment was in June (now, just four months away)!

Though it is only an hour drive (in good weather), it is a much more treacherous drive in winter because I would've had to drive up and down and incredibly winding, one lane road.

On August 2nd, I learned a couple of physician assistants were taking new patients in the next town over from mine, in Adams, MA. I am trying to switch to them to avoid the longer and dangerous drives to the other one I've been seeing since June.

It has truly been a stress-filled time for me. While I can speak on my own firsthand experiences in western MA, I've heard that this is going on all over the state . . . .and wouldn't be surprised if it's happening all over the country. For patients with MSUD and those that have been transplanted (as you can see from my experience), it is essential that we have a primary care doctor nearby. If I was sick, having to drive that long in bad weather would be very risky. Having someone who knows my health background (and is close by) makes the situation easier to bear. So, if you are facing a similar situation to mine, don't give up! Call around. Ask around. Hopefully you will get somewhere also.

MSUD RESEARCH UPDATE

By: Karen Dolins, Research Lead

These are exciting times for rare disease research. Advances in gene therapy for specific conditions make headlines, providing hope that gene therapy for MSUD will become a reality. Indeed, several researchers in the US and abroad are working on developing gene therapy techniques specific to MSUD. While it is exciting to know that scientists are hard at work and that technologies are evolving, challenges remain. The timeline needed to develop a new therapy is long and costs are high. Our research fund allows us to provide funds to initiate projects or to provide bridge funding until larger grants become available. Equally important, our support for a project demonstrates that friends and family affected by MSUD feel strongly that it is a worthwhile avenue of investigation.

Through generous donations, the MSUD Family Support Group has supported two gene therapy projects this year:

Muscle-directed gene therapy for MSUD, Institute Imagine, Paris, France: Our grant enabled post-doctoral student Juliette Lemoine to investigate the feasibility of a muscle-directed gene therapy for MSUD. Working under the guidance of renowned researchers Dr. Manuel Schiff and Dr. Clement Pontoizeau, Juliette's project explores an alternative to gene therapies targeting the liver. Most of the enzymes responsible for metabolizing the branched-chain amino acids (BCAAs) affected by MSUD are found in the muscle, making this an important avenue of investigation. Liver transplantation has taught us that providing a relatively small amount of functioning enzyme to the liver is adequate to normalize blood amino acid levels in most cases, but improving the ability of muscle to metabolize these amino acids could potentially provide even greater stability. The group in Paris along with their partner Généthon has designed a vehicle to deliver a transgene for the enzyme to muscle which they tested in MSUD mice. They were able to show improvement in survival and a correction of BCAAs.

Gene therapy in a bovine model of MSUD, University of Massachusetts: Dr. Kevin Strauss of the Clinic for Special Children has been collaborating with a team at the University of Massachusetts to develop a gene therapy that would be effective in a cow model of MSUD. MSUD occurs naturally in a line of Hereford cattle and has a physiological presentation that is very similar to that in humans. Along with veterinarian Dr. Heather Gray Edwards, they have successfully treated one cow who remains well 3 years after receiving a novel AAV gene therapy. The project requires the continued breeding and upkeep of this unique herd. We provided a grant to support the maintenance of the herd.

Additional ongoing gene therapy project: Dr. John Counsel at University College London continues his work developing a liver-directed gene therapy for MSUD. He and his colleagues have identified the most efficient elements to include in the final product and now are looking to determine the best dose to use in patients as they work towards a clinical trial.

Other projects:

• We continue to support the development of a home monitor for branched-chain amino acids.

• MyRareDiet: We are participating in a validation study for this app which will allow dietary intake to be logged and analyzed.

• Syntis Bio: Synt-203 is a product which will lower blood leucine levels by eliminating it from the gastro-intestinal tract. The company applied for a Small Business Innovation Research (SBIR) grant last year. The review has been slowed down by changes in Washington D.C. but does continue to progress.

• Examining brain function with fNIRS: Many attendees at the MSUD Symposium in 2024 participated in this study. The researchers have completed their data analysis and are now preparing a manuscript for publication. This work was supported by a grant from the MSUD Family Support Group Research Fund.

• MSUD patient registry with CoRDS: We are grateful to those who have participated in our registry as it allows us to learn about the lived experience of those with MSUD. We are currently analyzing the data and hope to publish our findings in a professional journal so that scientists, researchers, and clinicians can learn about the lived experience of those with MSUD.

There are over 1,000 rare diseases competing for funding and the attention of researchers. Please consider donating to the MSUD Family Support Group Research Fund so that we can continue to support worthwhile projects.

FROM THE PROFESSIONAL JOURNALS

Summarized by: Karen Dolins, Research Lead

Gene therapy in MSUD mice and cow

Dr. Jiaming Wang and colleagues at the University of Massachusetts describe the development and testing of their gene therapy construct for MSUD. They designed a recombinant adeno-associated virus (rAAV9) vector to deliver the genes for the 2 most common subunits affected by MSUD, BCKDHA and BCKDHB.

Importantly, while other techniques deliver the gene only to the liver, this team's product is directed to the liver, muscle, heart, and brain. This is critical as most of the enzyme affected by MSUD is found in the muscle, and the brain is also affected by the disease. They were able to show prolonged life, growth, and absence of neurological complications in both animal models.

Wang, J., Poskitt, L.E., Gallagher, J., Puffenberger, E.G., Wynn, R.M., Shishodia, G., Chuang, D.T., Beever, J., Hardin, D.L., Brigatti, K.W. and Baker, W.C., 2025. BCKDHA-BCKDHB digenic gene therapy restores metabolic homeostasis in two mouse models and a calf with classic maple syrup urine disease. Science Translational Medicine, 17(787), p.eads0539.

Thiamine-responsive maple syrup urine disease missed by newborn screen: A case report

Clinicians at Tulane University School of Medicine reported on the case of a child with thiamin-responsive MSUD that was not diagnosed at birth through newborn screening. She was referred to their clinic at 8 months of age when she failed to meet developmental guidelines. A workup including an amino acid analysis found significantly elevated branched-chain amino acids.

Upon admission, the child was placed on IV dextrose, large doses of thiamine (25 mg twice daily), and a metabolic formula free of branched-chain amino acids (BCAAs). Genetic analysis revealed a defect in the gene coding for the DBT subunit of the enzyme affected in MSUD.

At the time of the report, the child was 2 years 7 months old and was tolerating a diet with normal protein intake along with continued supplementation with therapeutic doses of thiamine. Isoleucine and valine supplements were no longer needed. She continues to take a small amount of metabolic formula, but the team is considering discontinuing its use as her intake improves.

This case study emphasizes the importance of early diagnosis. Thiamin-responsive MSUD is rare, and newborn screening may be conducted before BCAA levels become elevated. While the child is improving, developmental delays remain.

Upadia J, Noh G, Crivelly K, Smith J, Andersson HC. Thiamine-responsive maple syrup urine disease missed by newborn screen: A case report. Mol Genet Metab Rep. 2025 Aug 7;44:101244. doi: 10.1016/j.ymgmr.2025.101244. PMID: 40823510; PMCID: PMC12351175.

ABBOTT NUTRITION HEALTH INSTITUTE METABOLIC CONFERENCE 2025

By: Karen Dolins, Research Lead

Abbott Nutrition holds a conference for metabolic dietitians every other year. This year's conference was held in Columbus, Ohio. MSUD Family Support Group's Research Lead Dr. Karen Dolins was invited to present on the topic "Sports Nutrition and Inborn Errors of Metabolism: Providing Recommendations with Little Research to Guide Us."

A metabolic dietitian all-star cast presented a variety of other topics including "Navigating Nutrition for MSUD," "What the Heck Does that Laboratory Value Mean," "Introducing Food Allergens for Babies with Amino Acid Disorders," and more.

In addition to the conference sessions, the MSUD Family Support Group was represented by Board President, Sandy Bulcher, in the exhibit hall, allowing attendees to learn more about our disorder. There were approximately 100 attendees.

MSUD REPRESENTED AT THE INTERNATIONAL CONGRESS OF INBORN ERRORS OF METABOLISM (ICIEM) IN KYOTO, JAPAN

Researchers, clinicians, and patient advocates from around the globe travelled to Kyoto, Japan recently to attend the ICIEM conference and learn about advances in research and treatment. MSUD Family Support Group Research Lead Dr. Karen Dolins was one of 2,800 participants from 85 countries in attendance.

Topics were wide-ranging, including reports on advances in gene therapy and the current state of newborn screening. A session on "Challenges in Adult Metabolic Care" addressed the need for regular follow-ups and concerns related to body weight, exercise, and sexuality.

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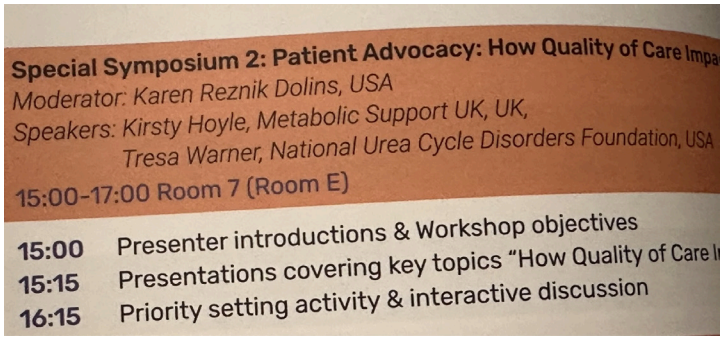

Karen moderated and presented at a Special Symposium entitled "Patient Advocacy: How Quality of Care Impacts Quality of Life." Joined by Tresa Warner of the National Urea Cycle Disorders Foundation (NUCDF) and Kirsty Hoyle of Metabolic Support UK, the trio discussed lifestyle issues including physical activity and how patient advocacy groups can assist clinicians in improved care and improved quality of life for those affected by inborn errors of metabolism (IEMs). All 3 speakers emphasized the important role of physical activity for overall health and wellbeing. While much research is needed, it was noted that people with IEMs are safely participating in sport and exercise and that it is important to include the medical team to monitor metabolic control while making changes in physical activity levels and to obtain recommendations for adjustments to diet.

Several posters and sessions addressed metabolic care for MSUD. Dr. Juan Francisco Cabello of the University of Chile discussed treatment and long-term follow up in that country. In Iran, where isoleucine and valine supplements are not available, Maryam Ziadlou described the efficacy of providing a formula which contains isoleucine and valine but not leucine (Xleu Maxamaid) during times of metabolic crisis. A poster described MSUD in Korea, where 22 patients have been identified over a span of 25 years. Newborn screening is now available in Korea, allowing early diagnosis and treatment. Other posters provided information on treatment and outcomes in India, the Philippines, Thailand, India, and Spain.

In addition to the sessions, Dr. Dolins was able to network with researchers, clinicians, and other patient advocates. With an estimated 1,000 plus inborn errors of metabolism, our goal is to encourage research which will improve the lives of those with MSUD.

ADULTING 101: FINDING YOUR PATH WITH HCU & MSUD

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

Growing up with classical Homocystinuria (HCU) or Maple Syrup Urine Disease (MSUD) comes with unique challenges—but no one has to face them alone. On September 11, 2025, the HCU Network America and the MSUD Family Support Group hosted Adulting 101: Find Your Path with HCU & MSUD, a free, empowering webinar designed for teens and young adults navigating the journey to independence.

The session drew about 30 attendees who tuned in to hear Dr. Jessica Gold, MD, PhD of Northwell Health, share practical insights about adulting with a rare disease. Dr. Gold's presentation focused on important aspects of managing life with HCU and MSUD, including self-management strategies, how to balance school, work, and daily routines, and the importance of staying on top of healthcare needs and medical appointments. She also highlighted approaches to building a fulfilling social life and maintaining healthy relationships, developing independent living and life skills, and planning for education, career, and long-term goals.

In addition to Dr. Gold's presentation, participants had the opportunity to ask questions and hear directly from community panelists living with HCU and MSUD, including:

- Amber Raye, Living with Classical MSUD
- Gabbi Lewis, Living with Classical HCU
- Jenna Mossner, MSW, LCSW, Living with Classical MSUD
- Ben Massengale, Living with Classical HCU

These panelists offered powerful personal stories and practical advice about balancing school, work, social life, and health responsibilities, while planning for the future.

Reflecting on the collaboration, Danae Bartke, Executive Director of HCU Network America, shared:

"Patients with inborn errors of protein metabolism often face overlapping needs. Our collaboration with the MSUD Family Support Group allows us to build perspective, pool expertise, tools, and allow patients to remain at the center of our missions. #StrongerTogether"

Denise L. Kolivoski, MBA, Executive Director of the MSUD Family Support Group, also emphasized the impact of the event:

"This webinar truly showed the power of collaboration. By working together with HCU Network America, we were able to bring two communities together, share experiences, and provide resources that empower young people to take charge of their health and their future."

The collaborative webinar underscored that while the path to adulthood with a rare metabolic disorder may be different, it is also filled with resilience, triumphs, and supportive communities.

Thank You to Our Sponsors
This important program was made possible through the generous support of Cambrooke, Nexus Patient Services, Nutricia, and Vitaflo. Their partnership ensures that the HCU and MSUD communities continue to have access to meaningful resources and programming that empower individuals and families to thrive.

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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?

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