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MSUD NEWSLETTER

Published by the MSUD Family Support Group ▪ Issue 42-1 ▪ Spring 2024

20TH MSUD SYMPOSIUM 2024: PRESENTED BY LEADING SPONSOR, NUTRICIA

The 20th MSUD Symposium 2024 is fast approaching, and we could not be more thrilled! It has been quite some time since we all gathered, making this conference particularly special. You will not want to miss the in-depth review of MSUD research. Numerous presenters will be sharing new research findings or providing updates on their ongoing projects through concise presentations. Alongside research, we will cover a range of other topics including medical and dietary management, liver transplantation, neuropsychology, advocacy, and fundraising. In addition to expanding your knowledge of MSUD, you will also have the chance to reconnect with old friends and meet new families. We're excited to announce a lineup of exceptional speakers for the symposium. Check out our complete list of speakers and agenda online at www.msud-support.org.

MSUD Symposium Hotel Reservations

Join us for our upcoming 20th MSUD Symposium at the Hyatt Regency in Reston, VA, from June 27 to 29, 2024. Conveniently situated near Dulles International Airport, this venue offers easy access for attendees coming from various locations. The proximity of the Metro Town Center station to the host hotel ensures convenient travel connections. Rooms are available with two double beds or one king bed for $159 per night plus tax. Reserve your room here: https://www.hyatt.com/en-US/group-booking/RESTO/G-MSUD.

MSUD Symposium Conference Reservations

Attend the entire three-day conference for just $30 per person or a maximum of $100 per family. This fee helps cover symposium costs. Payment can be made conveniently through PayPal by clicking the registration link here: https://msud-support.org/elementor-6896/. Simply fill out the registration form and click "Submit" to proceed to the PayPal site for payment. PayPal account holders can use their accounts, or guests can pay with a credit/debit card. Alternatively, payment by check is accepted. Make checks payable to "MSUD Family Support Group" and mail them along with the registration form to Denise Kolivoski, Executive Director- MSUD Family Support Group, 4028 Calico Dr, Erie, PA 16506.

MSUD Symposium Conference Meals

On Thursday evening, June 27th, during the registration/reception, dinner will be served, featuring regular and low-protein pizza, salad, and regular and low-protein cookies. Breakfast buffet will be available on Friday, June 28th, along with a lunch buffet on the same day. Additionally, a breakfast buffet will be provided on Saturday, June 29th for all attendees, with low-protein options available for all meals. Attendees are responsible for their own dinner on Friday evening, June 28th.

MSUD Symposium Support

Aside from research, the biennial symposium is the single most expensive activity funded by the MSUD Family support Group all year. Your donation is appreciated to ensure the continuation of this valuable and enriching experience for the MSUD community. Donate here: https://msud-support.org/donations-2/ Thank you for your generosity and compassion. ■

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FROM THE PRESIDENT'S DESK

By Sandy Bulcher

I hope that you are as excited about the MSUD Symposium 2024 as I am! I believe that you are, because I have been hearing from many of you, so I expect this symposium to have a great turnout. Start planning now to attend this event June 27-29, in Reston, VA at the Hyatt Regency. The hotel is located about 45 minutes from downtown Washington DC, which makes it an ideal location to tack on a family vacation.

Symposium details are available on the MSUD website www.msud-support.org. You can make your hotel reservation and pay your registration fee on the MSUD website, too. We have made it as affordable as possible to attend. The cost is $30 per person with a maximum of $100 per family. Check the website frequently for updates on the symposium.

With this big event comes a significant cost to the organization. In fact, it is our biggest expense of the year. If you would like to donate to help cover expenses, please reach out to our Executive Director, Denise Kolivoski, at execdirector@msud-support.org or 814-580-8449.

If you are unable to attend, check out the website after the symposium where you will find recordings of some of the presentations.

See you in June! ■

FROM THE DIRECTOR'S DEN

By Denise L. Kolivoski, MBA

Thank You for Making the MSUD Pet Pageant a Success!

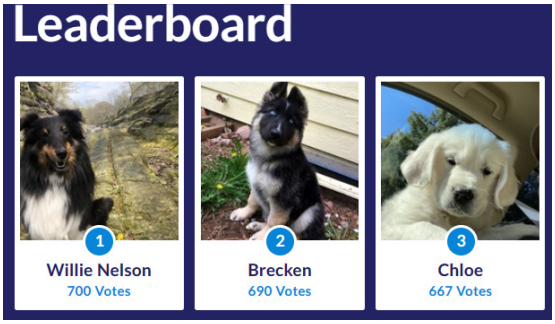
On behalf of the MSUD Family Support Group, we are excited to extend our heartfelt gratitude to each one of you for your enthusiastic participation in the first ever MSUD Pet Pageant. Together, we have achieved something truly remarkable.

Thanks to your dedication, support, and the overwhelming response from our community, we are thrilled to announce that we have raised over $6,000 for the MSUD Family Support Group. This significant contribution will help provide education, support, and advocacy to individuals and families affected by MSUD, allowing us to carry on our mission to make a positive impact.

Congratulations to Willie Nelson on being crowned the MSUD Pet Pageant 1st Place Champion!

We want to express our gratitude to everyone who voted, shared, and cheered on these amazing pets. Your collective efforts have made a real difference, and we are excited to share the great news that we are launching another Pet Pageant with even more fabulous prizes! Thank you once again for your incredible support. I look forward to your continued participation and the possibility of making an even greater impact with our next Pet Pageant.

Warm regards,
Denise L. Kolivoski, MBA
Executive Director
MSUD Family Support Group ■



FROM THE EDITOR'S LAPTOP

By Susan Needleman

Here we are already at our spring newsletter. I wanted to take this editorial to talk about a personal experience I have had over the past few years, with my formula. I was on the same formula since I was a toddler, until 2017, when I was 27 years old, it was discontinued. Like many of you have found yourself in recent years, I, too, was very worried about switching to something else and whether I would be able to find something I liked. I tried every formula on the market and different combinations of them. I finally found one, but it was unfortunately short lived. The formula I switched to did not agree with my stomach, while it did not make me throw up, it did not agree with me. After what seemed like a very long 18 months, I switched formulas again. I tasted a different formula that I had tried 18 months before and back then thought it was too strong. This time I added extra water and to my delight it tasted remarkably like the one I had grown up on. I happily stayed on this formula for two years, until another hurdle happened. Without warning the makeup of the powder I was putting in the formula changed, and I could no longer tolerate even a couple of sips of it.

Once again, I found myself in that worry of not knowing what I was going to do for a new formula and if I was going to find one. I retried a few that I had tried in the past few years, and one new formula had just gone on the market. Originally, I tried making it the same way I always had, with making enough in one batch to last all day. Something made me try making only half of the amount and, to my surprise, it tasted great! All I would have to do was divide it into two batches a day instead of making all my formula for one day at once and I would be all set, or so I thought.

Prior to switching this time, I was tolerating 600mg-720mg of leucine a day (10g-12g of protein), sometimes a little more, which had been an increase from what I was having years before. Suddenly my levels went up after switching formulas, which they hardly ever did, and I was now tolerating 120mg-180mg of leucine (2g-3g) of protein a day. No one knew why. I racked my brain, did some research, and found out it had to do with my formula. It had half of the amount of alanine, an amino acid that is not restricted for us. While this was an acceptable amount for someone with MSUD, my body got used to more from my prior formula. I did what needed to be done to get a prescription of alanine supplements to add to my formula. Over the next year, I kept slowly increasing the amount and I am happy to report that I am now back to my higher tolerance.

To learn more about Alanine, please read the Fall 2023 Newsletter (https://msud-support.org/wp-content/uploads/2023/11/volume-41-2_fall-2023.pdf), Alanine Supplementation in MSUD, on page 12. ■

FROM NIKOLAI'S WRITING NOOK

By Nikolai Rudd

While I have not been directly connected to a metabolic clinic since my transplant back in 2006, I have heard that some of the clinics have changed since I was, and I wanted to take the time to reflect on this.

Growing up, I was taken care of at Johns Hopkins University Hospital. Their new test detected my Classic MSUD for the first time in the state of Maryland. The Genetics Department was headed by Dr. Thaddeus Kelly and Dr. David Valle, over the next few decades, becoming one of the pioneering hospitals for MSUD on the east coast, (and) the country. After moving to Massachusetts in 1990, I switched to the care of Dr. Vivian Shih at Mass General Hospital. And because of her and her team of doctors, I was able to keep my leucine levels in the normal range, which for me was 100 umol/L - 125 umol/L, before electing to have the liver transplant for a better quality of life.

Recently, I learned that some MSUD adults feel that adult care is not as strong as care for younger individuals with MSUD. I am part of the first generation with it and there was so little known about MSUD at the time. Because of this, not much is known about how adulthood will affect our levels . . . and I haven't heard of any ways that they are figuring this out in a way that can benefit the adults living with it today.

Hopefully as more of us grow up, more can be discovered and the treatment for MSUD can be known at all ages. Moving forward, I hope that more people from Johns Hopkins University and Harvard University (which Mass General Hospital is connected with) as well as other medical schools and hospitals go into the field of Metabolic Research and find new treatments for MSUD moving into the future. ■

MY STORY: WHY I AM A DEDICATED FUNDRAISER FOR MSUD

Hello everyone! My name is Herb Foster, but everyone calls me Butch. My wife Diane and I are the parents of three children, two of which were born with classic MSUD – our son, Scott, and our daughter, Katelyn.

Scott was the first person in the Commonwealth of Massachusetts to be diagnosed with MSUD by newborn screening and would have been 53 this year had he not succumbed to a complication of MSUD, at the age of 22. He was one course away from completing his associate degree, which was bestowed upon him upon his passing. While on his college breaks, he interned at the Massachusetts Bay Transportation Authority (MBTA), where I was working as a safety official. His boss at the MBTA saw something in Scott and invited him to take the test to become a full-time employee. Scott passed the test and was hired as a motorman driving the Rapid Transit Trains on the Red Line. He was following in my footsteps as I had started in the same position on the same line years before.

After Scott passed away I wanted to do whatever I could to make a meaningful difference for children born with MSUD. Along with many friends and family, I started the Scott C. Foster Metabolic Research Fund. We started with an annual road race and at the first event we had well over 300 runners. My friends and I had gotten into running for exercise to stay in shape, so it was natural.

The very first contribution we made from the fund was to the Massachusetts General Hospital in Boston where Scott and Katie were treated. Shortly after establishing the two annual charity events, we started an annual golf tournament. We sponsored and managed these three annual events for ten years.

In the year 2000, I volunteered to host the biennial MSUD symposium in Boston. I was asked to raise $10,000 to support the event, which I gladly did. There also I met so many MSUD families and was totally moved by the many stories of parents' personal journeys with their MSUD children.

Several years ago, the MSUD Family Support Group learned of the Million Dollar Bike Ride run by the University of Pennsylvania Orphan Disease Center to support research efforts for orphan diseases. We applied to participate on behalf of MSUD. It was an easy decision because they were offering matching funds up to $50,000 the first year and then up to $30,000 each year thereafter. They put out the requests for proposals (RFP's) from scientists and administered the money we raised to successful applicants. We participated for five years and raised $324,698 over these years. I was so thankful that a group of MSUD "parents and patients rode each year, some in Philadelphia and others remotely.

This ended up being a godsend to me as I got back into biking and raising money for MSUD research again. Now that I am older and retired, biking has been a life saver for me, helping me stay healthy and in shape. Over these last five years I have raised an additional $75-$80,000 riding. This year, my 2 Gran Turismo biking partners and I plan on riding 77 miles in honor of my 77th birthday. I am just speculating, but my goal this year is to raise $25,000 and bring my total lifetime fundraising efforts on behalf of MSUD to $500,000 dollars.

The reason I am telling you about my experiences with fundraising is not for my own personal gratification! It is in the hope that it will inspire and stimulate all our MSUD families and friends to get on a bike or do whatever you feel comfortable doing to help the MSUD Family Support Group Research Fund. Many hands make light work! We as an organization have shown the ability to come together and raise money through the Million Dollar Bike Ride. Let us keep it going! The Board of Directors of the MSUD Family Support Group are on a mission to make lasting medical improvements to help all MSUD families everywhere. It has been a long journey for many families holding on to hope for their children. When my son Scott was born, they told us that in 10-20 years there would be a cure of some kind for his rare disease. Because our children were born with an extremely rare orphan disease, it is taking longer. The Board and I are encouraged by the interest from researchers in MSUD, and we are not sitting still in our efforts to help our families. We are being very proactive, but we need research funds to accomplish our mission.

Personally, I will keep riding my bike to raise research funds once a year until I cannot ride anymore. I am appealing to all families that raised funds for the Million Dollar Bike Ride in the past to get back on your bikes or come up with your own fundraising ideas! To all families reading this letter that hope and pray that someday there will be a cure for MSUD, you can make a difference. If every family did their part to raise just $1,000 a year, we can transform the landscape for MSUD. Our MSUD Board

'My Story' continued on page 9

IN MEMORY OF JESSE KIEL

By: Sandy Kiel

Mom to Jesse, 30 Years Old, Transplanted

Our dearly loved son, Jesse, died on September 18, 2023, at age 30, after a 320-day battle against Post-Transplant Lymphoproliferative Disorder (PTLD). Jesse was 30 years old and 11 years post-transplant.

Jesse was born April 12, 1993 – a healthy baby with MSUD. I had an amniocentesis, a test for chromosomal abnormalities and fetal infections, during my pregnancy because his sister Jenna was born two years earlier and she was detected by Michigan's newborn screening for MSUD. Newborn screening in Michigan began in 1987, and our two older children (non MSUD) were born before that. Jenna was the first baby in Michigan to be born since the screening had begun. So, Jesse had the best start to life – beginning immediately on MSUD formula. Jesse had a few childhood hospitalizations – the worst being at age eight when a flu virus caused brain swelling. But overall, he was a healthy, smart, and funny kid. He handled his MSUD diet well, drank his formula with gusto, and LOVED ketchup on everything - including his favorite potatoes, but also fruit and vegetables! He had a shirt to that said, "I Put Ketchup on My Ketchup". Many of you may know him from the MSUD Symposiums as we have made many dear friends within the MSUD community.

In 2007, Jenna was transplanted, but Jesse waited until he was 19 years old to choose transplantation in 2012. He had difficulty as a young adult in college and a couple of illnesses caused mental intoxication that was alarming. Jesse's transplant at Pittsburgh Children's Hospital was smooth – he had little complication and had since been able to take quite a low dose of immunosuppression. He had one round of rejection at two years out. He later transferred his care to the University of Michigan at age 26. Jesse was so thankful for his liver transplant – our kids say that after transplanting their mind was like the 'Claritin Clear' ads – finally clear-thinking and out of the clouds. Jesse completed his degree in Music Education and performed in the elite choirs at Grand Valley State University. He was a gifted singer, drummer, and was also involved in worship music. Jesse had a love of making unique drums. His favorite was an empty propane tank that had cuts in it to sound like a steel drum when played with his hands. He loved playing video - games with friends, especially Rocket League, board games, and had a Dungeons and Dragons group for which he made intricate playing pieces. Jesse was a Customer Service Representative at a West Michigan and manufacturer and was given the "Representative of the Year" award in 2023.

And on November 1, 2023, life changed . . . Jesse woke up with a pain in his side and we went to the ER in our town of Grand Rapids. He was diagnosed with a perforated bowel, and surgery was performed to repair it. The following day we learned that testing showed the cause was post-transplant lymphoproliferative disorder (PTLD) which involves the immune system and causes white blood cells called lymphocytes to multiply. The next ten plus months were a rollercoaster of antibody treatments, another surgery, sepsis from leaking bowels, chemo, more chemo, and all the antibiotics in the arsenal. He was moved to the University of Michigan hospital, to inpatient rehab, back to the local hospital, and home a few times. Overall, Jesse spent 214 days in the hospital – always with family and friends by his side and supported by many prayers from the community. We were told PTLD was treatable and always had hope that cancer would be cured and there would be healing. If you knew Jesse, you knew he was a 'gentle' soul – he endured so much with little complaint. He was well-loved by the nurses on the 5th floor. But the cancer became chemo-resistant, the bowels always leaked infection, and on Monday, September 18, surrounded by a roomful of family, friends, and all our love – Jesse left that destroyed body to meet his Savior. We know Jesse is healed in Heaven, but we miss him dearly here on earth: Carl and I, his precious girlfriend Virginia, his brother Haijo and Lacy, his sisters: Sara and Rick, Jenna, and Mark, two nieces and a nephew, and his close friends that spent many days by his side.

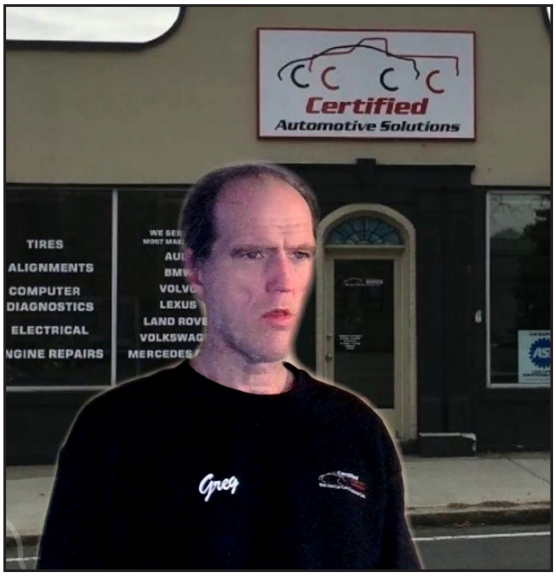
This is fresh for me to write, but our MSUD family is so dear to us. We have raised our children alongside many of you, reconnecting every two years at the Symposiums. We read the newsletters with great interest, and learned everything we could from other families and professionals, to do the best job we could to keep our children healthy. We all have this common link: having a child with this rare disease makes you learn to be an advocate for their care and wellbeing every day. What a long way MSUD knowledge, treatment, and transplant has come since 1991 when the providence of newborn screening saved our daughter's life and started us on this journey. If I have any bit of knowledge to pass on, I think it would be to emphasize always seeking out the best care for your children – be it with MSUD or transplant – and never stop being their advocate. We felt we made the best decisions at the time based on medical advice, but I will always wonder if we did enough. May God bless you and your children. ■

THRIVING AGAINST THE ODDS:

A LIFE WITH MSUD - THE STORY OF GREGORY WILSON

Interviewed By: Susan Needleman and Nikolai Rudd

Interviewee: Gregory Wilson, 47 Years Old, Classic MSUD



When parents find out their child has MSUD, most have not heard of it before and wonder what life will be like for their baby. Luckily, today, in many places, MSUD is picked up during newborn screening and doctors have a lot of knowledge of MSUD and can give adequate treatment, allowing their patients to have the most fulfilling life. This was the case in America in 1976, when MSUD had not even been discovered 20 years before. That is when Gregory Wilson was born and was one of these babies and one of the first to be picked up in newborn screening in Massachusetts. Little was known about what his future might be or if he had one, but he is still thriving to this day.

Recently, the editor and editor assistant of this newsletter, Susan Needleman, and Nikolai Rudd, got to sit down with Gregory and learn about his story.

His birth mother gave him up for adoption. He suspects his MSUD was part of the reason. He was adopted at six months old by a very caring and accommodating family. They monitored what he ate and that he took his formula, he never had to worry about it. His adopted sister even helped, by reading to him between sips of formula, motivating him to drink more. For this reason, he could not tell us much about the treatment for MSUD back then but knew if his parents were still with us that they would be able to.

Greg was still very much aware that he was different as a child. He was labeled and name called by his peers but was able to brush it off, he did not like these bullies anyway. He liked being different, as it would make things boring, if everyone was the same. He was also in Special Education (SPED) classes, and his classmates tended to stick together. He also got more attention in the classroom, which he enjoyed. He got taught extra tricks too, to help him remember concepts, which helped to a certain extent.

After high school he went to a local community college where he was in a SPED program that allowed him to earn his Associate's Degree in Business. He enjoyed this much more as he found himself interested in the concepts taught and could relate them to things in his own life.

As he became an older kid and later an adult, he began to recognize the presence of his MSUD more. This included noticing when his coordination and fine motor skills go, along with other symptoms. He has even set it up so he can test his coordination by walking in a straight line in his kitchen. He also finds that if he plays the game *Jenga*, he can determine if his levels are off, as it requires both fine motor skills and coordination. The game consists of pulling a block out of a tower of blocks and trying to avoid surrounding blocks from falling.

He went on to work in a mailroom in an engineering company for 20 years, switched to working as a Shipper at Marshell's Distribution Center, before changing his career to working in an automobile garage, where he cleans the shop and tests cars. To him, this is a big accomplishment as he always had a strong interest in cars, but as a child he was told he would never be able to drive or work, now he drives for a living! Due to his learning challenges, which probably are caused by MSUD, he is unable to, take the necessary courses to own his own classic car dealership, his dream job, but he is still thriving working, in a field he loves.

Greg might have started his life with an unknown future, but he has defeated the odds and now lives on his own and is working in his dream field. ■

MSUD Family Support Group Debuts National Radio Show Segment:

Amplifying Voices, Fostering Understanding

In a groundbreaking moment for the MSUD community, the MSUD Family Support Group aired its first national radio show segment on Tuesday, March 5th at 2:30 pm. Hosted by Dr. Marc Siegel of the Dr. Radio show, on SiriusXM Radio, channel 110, the segment provided a platform to shed light on the challenges faced by individuals and families affected by this rare metabolic disorder.

Dr. Siegel, known for its insightful discussions on various health topics, welcomed Sandy Bulcher, Board President of the MSUD Family Support Group, Dr. Karen Dolins, Research Lead and Board member of the MSUD Family Support Group and Dr. Vincent J. Carson, M.D., Pediatric Neurology expert from the Clinic for Special Children to share their experiences and insights.

Among them was Sandy Bulcher, the Board President of the MSUD Family Support Group who emphasized the significance of the occasion: "Being part of the conversation on Dr. Radio was an incredible opportunity to highlight the resilience of those affected by MSUD and the importance of comprehensive support networks in navigating this condition."

The discussion focused on the many challenges faced by individuals and families living with MSUD, from dietary restrictions to the emotional toll of managing a chronic condition. Through sharing personal stories and professional insights, the segment provided listeners with a deeper understanding of MSUD and the importance of support networks in coping with its complexities.

"Our discussion on Dr. Radio emphasized the significance of early detection, proper management, and ongoing research in improving outcomes for individuals living with MSUD," noted Sandy. "Every conversation about rare diseases brings us closer to advancements in care and support."

The MSUD Family Support Group, alongside Dr. Radio, highlighted the critical role of advocacy and community support in empowering those affected by MSUD. By raising awareness and sharing resources, they aim to ensure that individuals and families impacted by this rare disorder have access to the support and information they need to thrive. "The opportunity to collaborate with Dr. Radio enabled us to highlight the multifaceted challenges faced by individuals and families affected by MSUD," affirmed Sandy. "By raising awareness and sharing resources, we empower our community to navigate this journey with resilience and hope."

As the MSUD community celebrates this milestone, they look forward to continuing their efforts to raise awareness, advocate for research, and provide support to those affected by this rare disorder. Through initiatives like the national radio show segment, they hope to foster a more inclusive and understanding society for individuals living with MSUD and other rare diseases. ■

'My Story' continued from page 5

of Directors and our Executive Director, Denise Kolivoski, needs everyone's help.

When my son Scott passed away after coming down with the flu, our family was devastated. I thanked God that Scott had 20 healthy years and was able to lead a normal life with MSUD. I also thank God every day for giving my daughter Katie two beautiful children, an education, and the desire to give back and help children with special needs.

I firmly believe that God hand-picked each of us parents to raise our very special children. They are the ones that need our help every day. They are our heroes! Please help them. Together we can make a difference. God bless us all as we continue this long journey and pray that we will all see a very bright future for all children born with MSUD.

Sincerely,
Butch Foster, Director
MSUD Family Support Group
Call: (617) 875-6635
Email: scffund@comcast.net

P.S. – I will be riding again this year around the same time as the Million Dollar Bike Ride on behalf of MSUD research in early June. I encourage all families to participate in one way or another. For those who would like to participate and raise money or support my ride, donations can be made out to the MSUD Family Support Group/Research Fund. Feel free to contact me with any questions. ■

MAPLE SYRUP URINE DISEASE (MSUD) AND PREGNANCY

Amber Malik, MS, RDN, LD, Post Master Nutrition Fellow;
Rani H. Singh, PhD, RDN, LD Professor, Department of Human Genetics, Emory University

Managing Maple Syrup Disease during pregnancy demands meticulous dietary management to safeguard the health of both the mother and the developing baby. The primary focus of management is maintaining metabolic control while meeting the heightened nutritional demands across each trimester.

Individualized dietary guidance is important throughout pregnancy and the postpartum period. This involves precise adjustments in the diet, particularly protein intake from both medical foods and Intact protein (regular foods), while keeping the levels of branched-chain amino acids (BCAA) within the therapeutic ranges. This is key to addressing the unique nutritional requirements of pregnancy. As the pregnancy progresses the mother's ability to tolerate leucine increases, likely due to the fetus's metabolism of BCAA, allowing for greater consumption of intact protein [1,2]. The main goal is to keep BCAA levels as close to the normal plasma levels to ensure healthy fetal development. The recommended blood leucine (LEU) concentrations are 100-300 umol/L, with valine (VAL) and isoleucine (ISO) in the upper normal range of (200-400 umol/L) [1]. It is important to recognize that no teratogenic (detrimental to the fetus) effects have been found to be present in babies with women who had elevated plasma LEU levels during pregnancy, but it remains essential to deliver adequate nutrition to prevent maternal decompensation. [1]

Moreover, ensuring adequate energy at each trimester is crucial to avoid the risk of catabolism (breakdown of body tissue). Pregnant women with MSUD face additional challenges such as nausea and vomiting and have the added risk of catabolic crisis [1,3]. Preventing catabolism is a constant concern leading to the consideration of tube feeding or parenteral nutrition if oral intake is compromised due to appetite changes. During illness, combination of intact protein supplementation, with VAL and LEU, BCAA free medical food, tube feeding, and/or total parenteral nutrition (TPN) may be necessary [4,5]. Foods with modified low protein content can be incorporated into the diet to enhance variety and provide the additional calories necessary for supporting the pregnancy.

As the time for labor and delivery draws near, preparing to manage catabolism becomes critical, often involving IV glucose to prevent decompensation [5,6]. Postpartum period management is equally important to reduce the risk of metabolic issues, adjusting maternal leucine to pre-pregnancy levels unless breastfeeding [1,4]. Adequate energy intake remains a focus to prevent catabolism and sometimes necessary TPN [1,4,5,6].

The overarching goal during a MSUD pregnancy is maintaining metabolic control, with an emphasis on increased protein and energy intake, while meeting other nutrient needs to support the baby's development through each trimester and the broader demands of pregnancy [1,3]. This comprehensive approach aims to ensure the optimal health outcomes of both mother and baby throughout the MSUD pregnancy journey.

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CASE REPORT:

ACUTE ENCEPHALOPATHY IN A 10-YEAR-OLD PATIENT WITH MAPLE SYRUP URINE DISEASE: A CHALLENGING DIAGNOSIS

A recent report describes the treatment and outcome of two sisters living in Portugal with previously undiagnosed MSUD who presented to the hospital with acute encephalopathy during an influenza infection. The 10-year-old survived but her 18-year-old sister did not. Genetic testing revealed a variant of MSUD in both sisters. The author's note that MSUD has been included in newborn screening in Portugal since 2005, but that intermittent or intermediate forms may be missed making diagnosis difficult.

Miragaia P, Grangeia A, Rodrigues E, Sousa R, Ribeiro A. Acute Encephalopathy in a 10-Year-Old Patient With Maple Syrup Urine Disease: A Challenging Diagnosis. Cureus. 2024 Jan 27;16(1):e53043. doi: 10.7759/cureus.53043. PMID: 38410311; PMCID: PMC10895905. ■

ADVANCES IN GENE THERAPY FOR MSUD

By Karen Dolins, EdD, RDN

MSUD is caused by a defect in 1 of 3 genes: BCKDHA, BCKDHB, or DBT. While a defect in the BCKDHA or BCKDHB gene is the most common cause of MSUD, researchers have typically relied on a DBT mouse model with a less severe expression of MSUD for their studies.

Most of the enzyme needed to metabolize the branched-chain amino acids is found in muscle, but liver transplantation has taught us that only a small amount of enzyme activity is needed to normalize blood amino acid levels. This has led scientists to study the efficacy of liver-directed gene therapy.

In a new publication1, researchers at University Paris Cite in Paris, France reported the successful use of gene therapy to treat mice with Classic MSUD due to a defect in the BCKDHB gene. The same group previously developed a BCKDHA mouse model which they successfully treated using an adeno-associated virus (AAV) vector.2

Translating this work to humans will require much more work including a demonstration of safety.

1Pontoizeau C, Gaborit C, Tual N, et al. Successful treatment of severe MSUD in Bckdhb_/_ mice with neonatal AAV gene therapy. J Inherit Metab Dis. 2024;47(1):41-49. doi:10.1002/jimd.12604

2Pontoizeau C, Simon-Sola M, Gaborit C, et al. Neonatal gene therapy achieves sustained disease rescue of maple syrup urine disease in mice. Nat Commun. 2022;13:3278. ■

MSUD RESEARCH UPDATE

By Karen Dolins, EdD, RDN
MSUD FSG Research Lead

One way in which we attract researchers to MSUD is by attending professional meetings where we can exhibit and speak one-on-one with attendees and other exhibitors. The MSUD Family Support Group will be present at two professional meetings this spring: The Society for Inherited Metabolic Disorders (SIMD) and Genetic and Metabolic Dietitians International (GMDI) annual meetings which are being held back-to-back April 11-20 in Charlotte, North Carolina.

SIMD brings together clinicians and researchers with the aim of fostering research, increasing awareness, and improving treatments. Our presence there will enable us to connect with researchers with the goal of driving more research directed at improving treatments specific to MSUD.

GMDI is an organization of metabolic dietitians and allied professionals. Its mission is to provide standards of excellence in the nutritional treatment of inborn errors of metabolism. Our exhibit will enable us to provide participants with information about the MSUD Family Support Group including opportunities for research and education.

In addition to our exhibits, I will be part of a panel presentation at a joint session of SIMD and GMDI titled "Stronger together: The Case for Supporting Clinic–Patient Organization Collaboration." Other participants include representatives from flok (formerly PKU News), HCU Network America, the Propionic Acidemia Foundation, and 2 metabolic dietitians. The goal of the session is to improve collaboration between clinics and patient advocacy organizations thereby improving care. I will also be presenting a poster of our MSUD Registry research results at GMDI, enabling attendees to understand the gaps in knowledge which need to be filled.

The MSUD Family Support Group continues to monitor advances in gene therapy and other therapeutic modalities. We also continue to support the development of a home monitor for leucine. We are thrilled to be hosting an MSUD Scientific Update prior to our upcoming symposium. Members of our scientific advisory board and clinical advisory board will be present along with selected additional researchers as we discuss how to advance promising avenues in MSUD research. ■

CONSIDER LEAVING A LEGACY… A CHARITABLE BEQUEST

Imagine a world where individuals living with MSUD can experience more hope, support, and a brighter future. Your legacy gift to MSUD is the opportunity to turn that vision into a reality. By including MSUD in your estate planning, you are choosing to make a profound difference.

With your support, we can continue to fund vital research, raise awareness, and provide critical resources to individuals affected by MSUD throughout their lives. Charitable bequests are wonderful options for all donors, but especially those donors who wish to retain control of their assets during their lifetime.

How It Works

- Include a gift to the MSUD Family Support Group in your will or trust.
- Make your bequest unrestricted or direct it to a specific purpose.
- Indicate that you would like a percentage of the balance remaining in your estate or trust or indicate a specific amount.
- Tell us about your gift so we may celebrate your generosity now.

Benefits

- Your assets remain in your control during your lifetime.
- You can modify your gift to address changing circumstances.
- You can direct your gift to a specific area of interest in support of our mission.

Every legacy gift, no matter the size, plays a crucial role in shaping the future for countless families. Together, we can break barriers, find new treatments, and ultimately work towards a cure. If you are interested in learning more about leaving a legacy gift for the MSUD Family Support Group, please contact Denise L. Kolivoski, MBA. She can assist you through the process of setting up a bequest and help you in making a special gift to the MSUD community.

Contact Denise at execdirector@msud-support.org or call (814) 580-8449 to make your vision a reality. ■

SUPPORTING MSUD INDIVIDUALS AND FAMILIES: DONATE YOUR VEHICLE TODAY

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

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

Here is how it works:

1 Contact the MSUD Family Support Group to schedule a pickup or drop-off location for your vehicle.
2. Complete the necessary paperwork, including a donation form provided by the organization.
3. Receive a tax-deductible receipt for your donation.

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

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

ADVOCACY UPDATE

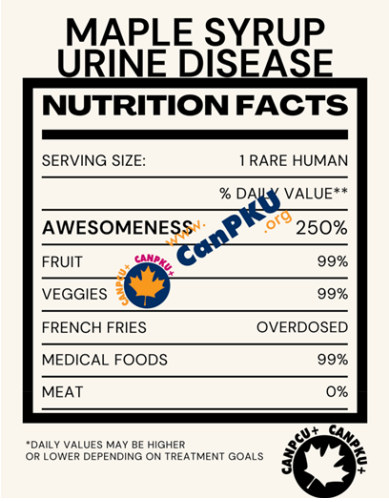
By: Jordann Coleman, Advocacy Chair

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

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

CanPKU+ wants to give you the opportunity to design what every you want with the fun MSUD image below! Pay just $5 (non-members) $4.50 (members) to download this image without the colored watermark. If inspired to increase the donation; we welcome this, simply adding more items to the cart increases the value of the donation. Once the purchase is complete, a PNG without the watermark will be emailed to the purchaser. At that point, the graphic can be applied to any product (Shirt, tumblers etc.) for PERSONAL - NON-COMMERCIAL use, by yourself or your favorite crafter!

Visit https://canpku.org/Purchase-CanPKU-Merch to purchase! ■



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 with the MSUD Family Support Group, please contribute a $50 gift today!**

You can show your support online at https://msud-support.org/donations-2 or send a check to:

MSUD Family Support Group care of Dave Bulcher, 958 Medinah Terrace, Columbus, Ohio, 43235. ■

GREAT OAKS
RETREAT CENTER

SAVE THE DATE
LINK COMING SOON

ILLINOIS LOW PROTEIN FAMILY CAMP

Check-in: 5 pm, August 9, 2024 | Check-out: 10 am, August 11, 2024
Great Oaks Retreat Center
1380 County Rd 900 N, Lacon, IL 61540

We are back this year! Join us back at the beautiful Great Oak Retreat Center for a weekend away with other PKU and IEM families. Memories will be made, meals will be shared and friendships will be fostered. There will be swimming in the lake, canoeing, paddle boating, arts and crafts, archery, high ropes course (ages 12+), ziplining (ages 12+), and fishing.

Friday night everyone is asked to bring their favorite low protein dish to share with the group - we will have a low protein potluck! All other meals will be included.

PRICE	RSVP BY
IEM Campers: Free Family Members: $10 each	July 19, 2024

a seriousfun camp

APPLY TODAY FOR THE SUMMER OF A LIFETIME!

Hole in the Wall offers a **FREE** week-long Summer Camp for children with serious illnesses aged 7 – 15. Campers participate in traditional camp activities while receiving onsite medical care from doctors and nurses!

SUMMER CAMP

- Bus transportation is provided from your clinic
- COVID vaccines NOT required

Apply for Summer Camp today!

FAMILY CAMP

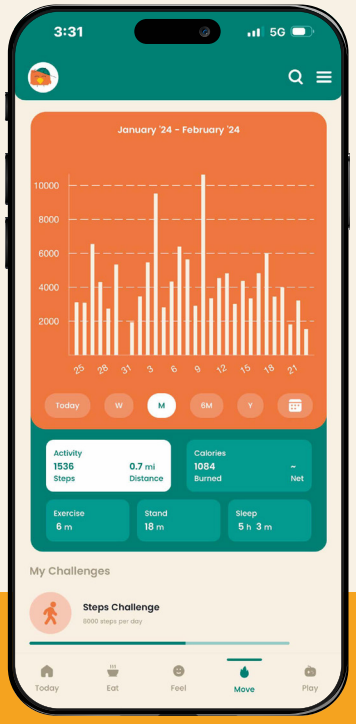
- Apply for a Spring Family Weekend!
- Diagnosed child aged 5-15
- COVID vaccines NOT required

Apply for a Family Weekend today!

CONTACT US

Camp: Admissions@holeinthewallgangorg | (860) 429-3444 | 565 Ashford Center Road, Ashford, CT 06278

flok Together: Camp and App with Us!



flok is advancing research and care for people with MSUD. **The flok app** helps you track how you eat, feel, move, and play to shed new light on our conditions.

Attend our in-person **flok Family Camps** in Oregon and New Hampshire! Our metabolic community members, family, and friends are all welcome for 3 days of fun, learning, and social connection.

flok FAMILY CAMP

Visit flok.org or call 943-777-3565 for more info on the flok app and Family Camp

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**

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

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

MSUD cooler®

- ✓ Suitable from 3 years of age
- ✓ Contains 15 g protein equivalent per pouch
- ✓ Available in Red and Orange flavors

Vitaflo™
Enhancing Lives Together
A Nestlé Health Science Company

Certified B Corporation

USE UNDER MEDICAL SUPERVISION. Be sure to check with your healthcare professional before making changes to your diet.

Let's Get Cooking!

MANGO SALSA SALAD

This flavorful recipe combines sweet and savory for a tangy dish that can be a filling entrée salad or a side dish for everyone at the table.

NUTRITION PER SERVING

Serving size: ½ of recipe
Calories: 170
Protein: 1.9 g
Phenylalanine: 72 mg
Leucine: 98 mg
Tyrosine: 33 mg

NUTRITION PER RECIPE

Makes: 2 servings
Calories: 340
Protein: 3.7 g
Phenylalanine: 143 mg
Leucine: 196 mg
Tyrosine: 66 mg

SCAN TO VIEW
RECIPE AND DIRECTIONS

For more recipes visit NutriciaMetabolics.com

NUTRICIA

Brought to you by Nutricia North America.

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

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WANT TO SHARE YOUR STORY?

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