MSUD
FAMILY SUPPORT GROUP

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MSUD NEWSLETTER

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MSUD REGISTRY: YOUR DATA PRESENTED AT NATIONAL AND INTERNATIONAL MEETINGS

By: Karen Dolins, Research Lead

The MSUD Registry is the most valuable tool available to our organization. Thank you to all who took the time to complete it. Your responses were presented to researchers, scientists, and clinicians at 2 meetings this past year, most recently at the Society for the Study of Inborn Errors of Metabolism (SSIEM) in Jerusalem, Israel. The information you provide will be used by researchers to develop research projects, clinicians to improve care, and the FDA as they evaluate new medicines.
The MSUD Registry will be most powerful if ALL questions are answered by a wide range of people. We continually evaluate your responses, allowing us to learn more about YOUR life with MSUD. It is also important to

'MSUD Registry' continued on page 4

2023 76 MILE CHARITY BIKE RIDE

By: Herb Foster

This year the MSUD Family Support Group did not participate in the U Penn Million Dollar Bike Ride, but I decided to continue my rides to raise much-needed money for our MSUD Research Fund. The Fund is named after my son Scott, who died of complications of MSUD at the age of 22.

Thanks to the Million Dollar Bike Ride I got back into bike riding to stay in shape.

With the help of my two friends, Billy Goodwin and Kevin Guinee, I was able to ride 76 miles this year and raise close to 15,000 dollars.

Next year, God willing, I will try and ride 77 miles for our research fund. I am so very grateful to all that contributed to this year's ride and look forward to next year's ride.

I will reach out to you all in the next newsletter to solicit your help and participation in next year's ride. Sincerely, Butch Foster, MSUD Board Member. ■

MSUD VOICE IS HEARD AT INTERNATIONAL MEETING

By: Karen Dolins

MSUD Family Support Group Research Lead Dr. Karen Dolins traveled to Jerusalem to participate in the 2023 SSIEM (Society for the Study of Inborn Errors of Metabolism) meeting which was held August 29-September 1. An estimated 1,400 scientists, clinicians, and industry members from around the world were in attendance.

Karen was part of an Advocacy panel which included Metabolic Support UK, Eurordis, and HCU (Homocystinuria Network America). MSUD families Shiri and Noi Widezki, Eden Zer, and Tal and Omer Brand were in attendance and participated in a discussion following the panel presentation, speaking about their experiences with MSUD in Israel.

This was an important opportunity for the MSUD Family Support Group.

'MSUD VOICE' continued on page 4

Inside This Issue:

Sections

Save the Date

MSUD Symposium 2024 will be held at the Hyatt Regency in Reston, VA on June 27th-29th. More information to follow. Plan now to attend!

FROM THE PRESIDENT'S DESK

By Sandy Bulcher

I can't believe that summer has come to an end. The MSUD Family Support Group has been busy this spring and summer. In May, I traveled to Washington DC and looked at 3 hotels for the MSUD Symposium 2024. We decided that the Hyatt Regency Reston, VA was the best fit. It is a beautiful facility for our event which will be held on June 27-29, 2024. Mark your calendars now to attend!

In June, we hosted a webinar to introduce our new executive director, Denise Kolivoski. She is our first paid employee and is working 20 hours per week. (see more about Denise in this newsletter). We're excited to have her onboard and expect great things!

Also in June, my husband Dave and I traveled to Erie, PA and met with Denise. We provided her with MSUD materials and education and reviewed her goals. She is learning quickly about MSUD and the needs of our small community.

The MSUD Family Support Group Board members and Denise met in Columbus, OH in July for our annual in-person meeting. During the two-day meeting, we covered many topics including plans for the next symposium. We also reviewed our research projects, communication tools, budget, and finances to name a few.

This fall, we'll be focusing on the symposium agenda. The 2024 symposium will be our first symposium since we gathered in Pittsburgh in 2018. I'm anxious to see everyone again and meet the new families! We plan to include speakers/topics that are of interest to all, including the transplant community. We hope to see many of you there!

We'll also be focusing on our fundraising efforts this fall and winter. As always, we need your help financially to continue our work. Donations can be made via check to Dave Bulcher, Treasurer (see more information in this newsletter) or online via our website www.msud-support.org.

On a personal note, our 33-year-old son, Jordan (with Classic MSUD), had his appendix taken out this summer. Thankfully his surgery was done at Nationwide Children's Hospital in Columbus, OH where his metabolic team is located. The surgery itself and the recovery from surgery went smoothly, but he had issues keeping his leucine from elevating despite several readmissions for IV fluids. One week after surgery, he was feeling well, and his leucine level returned to normal. We have much to be thankful for. There is little information about the effects of surgery on MSUD, so he'll be documenting his experience in the registry. The importance of collecting data via the registry to better understand MSUD cannot be emphasized enough!

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

FROM THE EDITOR'S LAPTOP

By Susan Needleman

It is hard to believe it is time to write my Editor's article again, but at the same time so much has happened since the last issue of the newsletter. As you may have heard (or can read about on page 3,) the MSUD Family Support Group now has its first Executive Director!!! I would like to officially welcome Denise Kolivoski to the MSUD Family! She has already brought a lot of great ideas to our group. Throughout this newsletter you will already see her impact on our organization, including a number of fundraising campaigns.

Fundraising is critical to achieving the mission of the MSUD Family Support Group. A big part of what we do is support research to improve the lives of all of us with MSUD and of course hold symposiums, which are coming back next year after a COVID hiatus. All of this would not be possible without your help. That is why, as an adult with MSUD, I would like to give a personal plea to everyone who reads this to please donate to our important group what you can. Your donations can help improve the medical care that I, and others with MSUD receive. Want to submit an article for the newsletter? Email my team at msudeditor@gmail.com or call 781-420-2676. ■

A MESSAGE FROM THE NEW EXECUTIVE DIRECTOR OF MSUD

Dear Members of the MSUD Community,

It is with great honor that I reach out to you today as the newly appointed Executive Director of the MSUD Family Support Group. As I step into this role, I am deeply humbled by the opportunity to serve and lead this incredible organization, which has been a beacon of hope and support for countless individuals and families facing metabolic disorders. With a background in nonprofit management, I'm excited to bring my experience in raising awareness and funds that can improve the quality of life for individuals and families living with MSUD.

As we move forward together, my vision is to strengthen the mission of our organization through education, research, and advocacy, creating a brighter future for all!

1. **Education: Knowledge is the cornerstone of empowerment.** We will strive to provide accessible, up-to-date, and accurate information about MSUD, its management, and the resources available to our community. Through educational initiatives, webinars, workshops, and partnerships with medical professionals, we aim to equip families with the tools they need to navigate their MSUD journey confidently.
2. **Research: Advancements in science and medicine hold the key to improving the lives of those living with MSUD**. We will collaborate with researchers, institutions, and organizations to support cutting-edge research initiatives, raise awareness, and foster innovation in treatment and care. By investing in research, we can pave the way for a brighter and healthier future for our community.
3. **Advocacy: Our collective voice is powerful, and together, we can drive change**. Advocacy will be at the forefront of our efforts as we seek to raise awareness about MSUD in society, advocate for policies that support individuals with metabolic disorders, and ensure access to comprehensive care and support services. Our advocacy efforts will be founded on empathy, compassion, and the conviction that every individual's voice matters.

I am eager to work alongside our exceptional team of volunteers, partners, and supporters, all of whom share the same passion and dedication to our mission. Together, as a united MSUD family, we will make a positive impact on the lives of people living with MSUD throughout the world.

Sincerely,
Denise L. Kolivoski, MBA
Executive Director, MSUD • execdirector@msud-support.org • 814-580-8449. ■

FROM EDITOR'S ASSISTANT'S WRITING NOOK

By Nikolai Rudd

When I first heard that the next MSUD symposium was going to be in either Baltimore, MD or Washington DC, I was elated! Even before the board finally chose Reston, VA as the final location, right near DC I busily started to make plans. (See page 1 for the announcement about the 2024 MSUD Symposium).

You see, I was born at Georgetown University Hospital in Washington, DC, and my MSUD doctors were at Johns Hopkins University Hospital in Baltimore, MD. My family and I lived there for the first 16 years of my life, but we moved up to the Berkshires of western Massachusetts in 1990 (the summer before my Sophomore Year of high school). And since moving . . . I don't get to visit DC nearly as much as I used to in the first couple years. And have only managed to see my old friends on rare occasions over the many years.

SO—as you can imagine—having the symposium down there is going to be a virtual "Home Coming" for me!! And a chance to see the changes of my old stomping grounds. Seeing my old haunts and trying to find places that were sentimental to me, that might still be around.

And this "Homecoming" of sorts would enable me to, not only, reconnect with all my MSUD and Transplanted friends, but also to spend a day or two longer so I can see some of my dearest and oldest friends whom I haven't seen in ages. And get a chance to hang out (in person), reconnect with them, and learn about each other in the present . . . while still reminiscing about ALL of our memorable times from the past.

I am also hoping that I will be able to see a couple of my old teachers who had a great impact on me when I was a student and also when I started teaching myself.

I am SO excited and am looking forward to seeing everyone next summer. And I'm sure that many others are just as excited. I hope to see you there! ■

'MSUD Registry' from page 1

update your responses every year as this allows us to see how your experiences change over time.

We plan to present a new analysis to the medical community in 2024, so if you haven't yet completed the questionnaire or provided your annual update, please do this soon!

New participants: Please take the time to register by going to our website: www.msud-support.org or http://research.sanfordhealth.org/rare-disease-registry.

Already registered? Email cords@sanfordhealth.org to update your information.

Not sure how to answer some questions? Contact Karen Dolins at 914-391-2982 or karen.dolins@gmail.com.

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

The importance of this registry to all with MSUD cannot be overstated. Completing the MSUD Registry is one thing you can do **TODAY** to improve the lives of those with MSUD and their families. Remember, all your information is de-identified so your responses cannot be linked to your name or other identifying information.

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

'MSUD Voice' from page 1

In addition to presenting on the lived experience for those with MSUD and their families where she cited data from our MSUD Registry and the work of Dr. Jessica Gold, Karen connected with researchers and industry members, emphasizing the needs of our community. She was able to share her ideas for future research priorities for MSUD which include the development of a monitor to measure blood leucine levels in real time, therapeutic agents to improve control of blood leucine levels, and gene therapy.

After the conclusion of the meeting she traveled to visit other MSUD families, met with the Chief Scientific Officer of an Israeli biotech company working on a potential therapy for MSUD, and met with metabolic physician Dr. Orna Staretz- Chacham and Dr. Lital Alfonta who are collaborating on a project to develop a leucine monitor (see article on page 15 for more information about this project). Our support group has provided funding for this project through the University of Pennsylvania's Million Dollar Bike Ride. ■

Gally and Yaron Peled

Dr. Orna Starertz-Chacham and Dr. Lital Alfonta and students

Star Family

ADVOCACY UPDATE

By Jordann Coleman, Advocacy Chair

In 2023, there is a crucial piece of legislation set to impact the MSUD community. The Medical Nutrition Equity Act (MNEA) stands at the forefront of this legislative change, 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 and subsequently re-introduced during the 117th Session of Congress in 2021. The previous iteration garnered significant support from various members of Congress, and now, the updated version is poised for re-introduction in the coming months in the 118th Session of Congress.

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

EMORY METABOLIC CAMP

By: Jean Handler, 38 Years Old, Classic MSUD, Transplanted

The Genetics Clinic at Emory University in Atlanta, Georgia, runs a week- long camp every year for MSUD and PKU females aged 12 and up called "Metabolic Camp". Every year my parents donate and also give a scholarship for campers who can't afford to go. My mom gave metabolic dietitian/researcher/professor, (I like to call her "Superwoman") Dr. Singh, the idea for a camp for girls who have metabolic genetic diseases around three decades ago. At the time I was a child, I wanted to experience a sleep away camp, like my classmates did. Thanks to this camp I was able to do so. I am one of the original campers and have been going for 29 years. My role now is that of a mentor. I will continue going until the day I die.

Camp means everything to me. It was different the last two years because it was held online. I rather it be in person as that way you get the true camp experience and the sisterhood that develops as we bond for life. This was our 29th year, you heard me 29 years of fun, dance parties, great memories learning new things about yourselves and how to maintain our special metabolic diets and prescribed formula and still live a happy life. Yes, I describe this particular camp not just as a camp, but as a sisterhood and longtime bonding experience of friends. All of the girls have faced a lot of challenges in their lives from handling their medical diseases to learning how to explain it to friends, peers, teachers, and other doctors that aren't familiar with MSUD and PKU and similar conditions. This year we learned four important goals: advocacy: loving yourself, being strong and brave, and being a survivor and a fighter and not a victim. Again, let me summarize this camp. It's not just a camp for me personally, it is my second family, my home away from home, my week where I can be myself and see all my great friends/metabolic sisters. Over the years I have become a leader, role model and a true friend who is always there whenever one of my camp sisters needs me and helps them to the best of my ability.

I've learned this year that it is important for everyone to stand up for yourself and fight for yourself, by advocating for everything needed to express yourself especially when people are new in your life. For example, if you're going to a doctor you've never been to before, you need to speak up about your disease, and what you're feeling, and what's happening with you. I felt that Camp 2023 was my year to learn how to grow and stand up for myself and be the best I can be and focus on my health and get into better shape so I can feel healthy physically, mentally, and at peace with myself and the person I have become - an independent, strong woman.

I have accepted that I am not a victim, I am a survivor. Even though I have this rare metabolic disease I won't let it define me or take over my life. I am much stronger than that and won't let anyone or anything ever defeat me. It was wonderful to see all the original campers and looking back at what we all went through in our lives, we are still stronger and better than ever.

To find out more about Emory University's Metabolic Camp visit https://metcamp.net/ ■

TEEN/ADULT VIRTUAL MEET-UPS

By Susan Needleman

There are a lot of people in this world, but few who have MSUD, or even know what it is. With all that is needed to care for MSUD, and all that is going on in the world now, it is more important than ever to have people in our lives who "get it". That is why I started MSUD Virtual Meet-Ups. It has really turned into a support group of friends. During these meetings we talk about MSUD and non-MSUD topics while we laugh, cry, or just listen. Teens and adults with MSUD are invited to join us on occasional Saturday nights at 8pm EST/ 7pm CST on Zoom. At these meetings teens and adults will be welcomed by a friendly group of others like them who have MSUD. If you would like to attend please email me at susanneedleman.msud@gmail.com.

DEBUTING MSUD'S PET PAGEANT

Flex your fundraising "MUSCLES" and help children and adults worldwide feel "MIGHTY" as they battle MSUD! Join the MSUD Family Support Group as we debut our 2023 Pet Pageant Photo Calendar Contest by submitting a photo of your favorite pet and vote for your favorite photo. If you don't have a pet, you can vote for someone else's pet to help them win the contest!

When you vote in or enter our 2023 Pet Pageant Photo Calendar Contest, each dollar you donate goes back towards helping improve the lives of children and families living with MSUD. Submit your favorite picture of your pet or pets for a chance to win amazing prizes that include gift cards to pet stores which you can use to pamper your pet!

The winning pictures will be included in our 2024 photo calendar and the three pets that generate the most votes win prizes. If your pet is a top vote-getter, you will receive one complimentary copy of the calendar which will be printed in a 12" x 24" format. Winners will be notified during the first week of January 2024.

All pets are welcome! Don't forget about your favorite pet fish, frog, or turtle. Every pet is eligible for an entry in MSUD's Pet Pageant. Each vote helps change lives. There is NO FEE to participate in this year's photo calendar contest! You can choose to donate $1 per vote to support your favorite pet or another pet that you think should win the top prize. You can vote as many times as you like with each $1 vote donated to MSUD Family Support Group.

Get ready to enter your favorite pet in MSUD's Pet Pageant! Mark your calendars - this contest kicks off on November 24. Join the fun at http://www.gogophotocontest.com/petpageant! ■

MY MSUD STORY

By : DeAuzjhae Mekhia Williams , 17 years old
Classic MSUD, Syracuse, New York

I was diagnosed with MSUD when I was a baby. The doctors did not know what was wrong with me and it took a week to diagnose me with classic MSUD. After the doctors did, they told my mom all about MSUD and how to manage it. MSUD is a rare medical condition when your body cannot break down protein. The treatments for MSUD are either managing a very restrictive diet to keep my levels under control, or having a Liver Transplant. Since, Liver Transplants are not always 100% effective, I have not had one and live on a strict low-protein diet. I can only eat fruits and vegetables and low protein foods and I drink this special formula to keep me alive. Throughout my whole childhood, I was in and out of hospitals. My mom made my formula, but she always told me that I have to learn how to make it. First I had to learn how much water I had to fill up in my container, then I poured the powder in and lastly, I learned the measurements and how to make it.

I didn't know how to make friends so I was by myself at school, and I was bullied because people misunderstood me. Some people even pitied me. Respect is very important to me because of how much I was bullied and people were mean to me. Although I did have friends, I didn't tell them about my MSUD. When I tried, they didn't understand, so I decided to be alone. I got comfortable with being that way. It took me a while before I felt comfortable talking to people and with dating. I felt if I told them about me, or showed any emotions, they would use that against me. So for a while I decided to focus on myself. I needed to learn how to love myself. MSUD made me stronger and it made me more independent. It also made me want to become a doctor. I want to be a doctor that cures rare diseases. I want to have a clinic called, "The MSUD Clinic." No matter how hard it gets, or how alone I feel, I'm going to be the best doctor I can be. MSUD made me realize that being normal is boring. Your difference Is what makes you who you are. ■

HAPPY 60TH BIRTHDAY TO DARRELL DEEL

Interviewed By: Susan Needleman and Nikolai Rudd

Interviewee: Bonnie Deel, Mom to Darrell, 60, Classic MSUD, Virginia

This October marks a momentous milestone in the MSUD Community. One of our oldest members, Darrell Deel, is turning 60 years old on October 12th! Happy Birthday Darrell! To mark this occasion our editing team (composed of Susan Needleman and Nikolai Rudd) interviewed his mother, Bonnie Deel, and found out about his amazing story.

MSUD was first described in 1954. The cause was identified in 1959. The oldest person living with MSUD was born in 1959. This meant that in 1963, when Darrell Deel was born in Freemount, OH, VERY little was known about the disorder. Much less than is known today. There was a very small number of people who had been diagnosed with it. Of those, many would end up passing away during childhood due to brain edema. Darrell's family was not going to have that be his destiny.

At the time, New York had more experience than Ohio with MSUD, as they recently had another baby with it. Darrell was sent over there for 9 months while the doctors studied MSUD and how to control it to learn about MSUD, and how to care for him. Every day they would send photos of him to his parents who had returned home to care for the rest of their family. His story quickly spread during this time and newspapers came to the hospital to take pictures of him and get his story. His follow-up care was in Ohio. Even after the family moved to Virginia, they would make regular trips to Cleveland, for many years.

Darrell has done very well from the care he received from his mother and his entire family. He did acquire diabetes and hearing loss, but no connection to these and MSUD is known. He still resides in Virginia to this day, with his mother, where he keeps himself busy in the house, and is fairly independent. He enjoys video games, watching movies, sometimes mowing the lawn, and is learning to cook. Happy 60th Birthday Darrell! ■

MSUD FAMILY SUPPORT GROUP INTRODUCES NEW SHIRTS

Show Your Support and Make a Difference!

We are thrilled to announce the launch of our brand-new MSUD Family Support Group shirts, featuring the inspiring slogan "I am Mighty with MSUD" and "Our Family is Mighty with MSUD." These shirts are not just a stylish addition to your wardrobe, but also a powerful way to show your support for individuals and families affected by MSUD.

Shirts are now available for purchase online at www.msud-support.org/shirts, making it convenient for you to get your hands on one and spread awareness about this rare metabolic disorder. There is also MSUD logo apparel such as golf polos available for purchase at www.msud-support.org/shirts. With the holiday season fast approaching, these shirts also make thoughtful gifts, allowing you to share the message of support and solidarity with your loved ones.

There is also international delivery of these shirts to most countries. Typically, the cost is US $20.00 or more and takes 3-4 weeks for delivery. You can call +1 703-434-3214 to learn about delivery options and what products are available in your country!

For those of you attending the 2024 MSUD Symposium in June at the Hyatt Regency in Reston, VA, these shirts will be available for purchase in-person. It's the perfect opportunity to grab a shirt, connect with fellow MSUD advocates, and celebrate our united efforts to make a difference in the lives of those affected by this condition.

The best part is that all proceeds from the sale of these shirts will directly benefit the ongoing initiatives and mission of the MSUD Family Support Group. Your purchase will contribute to our efforts, enabling us to continue offering essential resources, promoting awareness, and making a positive impact on the MSUD community.

Visit www.msud-support.org/shirts to make your purchase today or mark your calendars for the 2024 MSUD Symposium to buy the shirt in-person. If you would like to be a part of the "Mighty with MSUD" marketing campaign, please take a photo of yourself in the shirt showing your best muscle pose and email it to execdirector@msud-support.org to be a part of our future social media and outreach efforts!

Thank you for being a champion of MSUD and joining us in making a difference! ■

CONSIDER LEAVING A LEGACY…

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. Your legacy will be one of compassion, empathy, and belief in a world where MSUD no longer holds people back from living their fullest lives.

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 execdirector@msud-support.org or call (814) 580-8449 to discuss how we can work together to make this vision a reality. ■

ANNUAL DONATION REQUEST:

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

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

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

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

☐ $50 ☐ $100 ☐ $250 ☐ $Other _____

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

☐ Please remove me from the mailing list.

☐ I do not have access to view online newsletters and prefer printed newletters. (Spring newsletters are online only.)

☐ Please contact me about making a multi-year pledge and/or estate planning strategies.

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

Name | Professional Title (if applicable) | Telephone

Mailing Address | City | State | Zip | Country | E-mail

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

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

HELP THE MSUD COMMUNITY WITH A FACEBOOK BIRTHDAY FUNDRAISER

Birthdays are a time of celebration, love, and joy. This year, we invite you to add an extra touch of meaning to your special day by supporting the MSUD Family Support Group. By creating a Facebook fundraiser for your birthday, you can make a real impact in spreading awareness and raising funds to further our mission of education, research, and advocacy for individuals and families living with MSUD.

How to Get Started:

1. **Connect with MSUD Family Support Group:** To kickstart your birthday fundraiser, search for "MSUD Community Support" on Facebook and connect with our official organization page. By linking your fundraiser to our page, you'll ensure that all funds raised go directly to supporting our important work.
2. **Set Your Fundraising Goal:** Whether it's $100, $500, or any amount you're comfortable with, every contribution counts. Your birthday fundraiser's success is not just measured by the funds raised, but also by the awareness you can spread.
3. **Personalize Your Message:** Share your story and connection to MSUD with your friends and family. Let them know why supporting the MSUD Family Support Group is important to you and how their contributions can make a difference in the lives of those affected by this rare metabolic disorder.

Why Your Birthday Fundraiser Matters:

- **Raising Awareness:** MSUD remains a lesser-known condition, and your birthday fundraiser can help shed light on this rare disease. As you share information about MSUD and our organization's efforts, you contribute to raising awareness and understanding among your social circle.
- **Funding for Mission:** The funds raised through your birthday fundraiser will directly support our mission of providing education, research, and advocacy for MSUD. These resources are vital in enhancing the quality of life for individuals and families facing the challenges of MSUD.
- **Community Strength:** When you create a birthday fundraiser, you encourage others to engage and support the cause. By coming together as a united community, we can drive change and make a meaningful impact.

Spread the Word:

Use the power of social media to reach a wider audience. Post updates about your birthday fundraiser regularly, expressing gratitude to donors, and sharing MSUD-related facts and stories. You can find facts and information about MSUD on our homepage, https://msud-support.org/. Encourage your friends to share your posts, amplifying the reach and impact of your campaign.

Let's Celebrate Life Together:

Your birthday is not just a celebration of another year around the sun; it's a celebration of life and the joy of making a difference. By creating a Facebook fundraiser for the MSUD Family Support Group, you are turning your special day into an opportunity to help others, spread awareness, and supporting a community that needs our collective strength.

Remember, even the smallest gesture can have a profound impact on someone's life. Let's create a wave of change together! ■

Nexus Patient Services

Commited to making life a little easier for people with metabolic disorders through innovation, quality and customer service.

MSUD EASY TABLETS

Ask your dietitian for a sample today!

Easy To Measure ✓ — 11 MSUD Easy Tablets contain 10g protein equivalent

Easy To Take ✓ — Almond shaped tablets allow for easier swallowing with little to no taste or smell

Easy On The Go ✓ — Convenient to take while traveling, at work, or at school

MSUD Easy Tablets are coated slow-release tablets which are to be swallowed whole with liquid.

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VitaStories is a program designed to share your experience using **Vitaflo** formulas and discover firsthand how finding the right formula can make a difference.

Your story may be shared with the rare disease community to inspire, support, comfort, or motivate those with similar conditions.

Visit **VitafloUSA.com** for more information and resources.

To share your **VitaStory** or learn more about the program, scan the QR code or email: **VitaStories@VitafloUSA.com**

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ALANINE SUPPLEMENTATION IN MSUD

By: Dr. Jessica Gold
Cohen Children's Hospital, Northwell Health

Alanine is a neutral amino acid. Unlike the branched chain amino acids (isoleucine, leucine, and valine) which are essential (must be provided by the diet), alanine can be made by your body and is therefore non-essential. The branched chain amino acids can be used to make alanine. However, if your body cannot break down branched chain amino acids well, such as in MSUD, it may also have trouble making alanine. Branched chain amino acid metabolism is connected to alanine metabolism through the Glutamate-Glutamine cycle, which also has an important role in neurotransmitter synthesis. There are no known symptoms to having an alanine level that is too high or too low.

Alanine is important because of its role as a building block for proteins and as a source of pyruvate, an important source of energy. When diagnosed with MSUD, people tend to have very low alanine. These concentrations of alanine increase after starting an appropriate diet, but never entirely normalize. Daily alanine supplementation may help maintain the body's necessary amount of alanine and balance branched chain amino acid metabolism.

No formal studies have been performed on the benefits and risks of alanine supplementation in MSUD, but clinician experience has suggested that it may be helpful. At the Children's Hospital of Philadelphia, alanine supplementation is used as an additional therapy to the more common treatments of branched chain amino acid restriction and valine and isoleucine supplementation. We have found alanine supplementation to be especially helpful for individuals who have difficulty controlling their leucine levels and have low alanine on their plasma amino acid quantification. Alanine dosing starts at 150mg/kg per day, which is then divided into doses given 2-4 times daily, depending on patient preference. Following initiation, dosing is titrated based on alanine levels. Alanine can also be used as an adjunctive therapy to lower leucine levels during metabolic crisis. For patients in crisis who are already on supplemental alanine, dosing should be increased between 2-4 times their normal dose, similar to how valine and isoleucine doses are doubled.

It is important that patients speak to their metabolic doctor and/or dietitian regarding alanine supplementation before adding it to their regimen.

More information about the relationship between alanine and leucine can be found in these studies or you can ask your metabolic specialist.

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Yudkoff M. Interactions in the Metabolism of Glutamate and the Branched-Chain Amino Acids and Ketoacids in the CNS. Neurochem Res. 2017 Jan;42(1):10-18. doi: 10.1007/s11064-016-2057-z. Epub 2016 Oct 1. PMID: 27696119; PMCID: PMC5285401. ■

PKU NEWS BECOMES flok, EXPANDING TO SERVE PKU, MSUD, AND MORE

By: Sarah Chamberlin flok, Executive Director

Say hello to flok—the evolution of PKU News. This reimagined organization is deeply committed to uniting the inherited metabolic disorder community. Our aim is to continually elevate the standard of care and accelerate scientific research for our entire community. One of flok's first new offerings is a brand-new mobile app, also named flok, designed for those with PKU, MSUD, HCU, TYR, UCDs and Organic Acidemias.

We all understand that living with a metabolic disorder goes beyond managing diet. That's where flok comes in. The flok app helps you understand how you eat, feel, move, and play, and how all that affects your health. The tools it offers can enhance communication with your clinicians and foster a sense of community among users.

But that's not all—the flok app, which will be free to download and use, facilitates a community-driven approach to research. By incorporating your lived experiences, we'll be able to prioritize areas of research that matter the most to you.

Interested in being one of the first in the MSUD community to test the flok app? Our beta program kicks off this fall! To get involved, sign up at flok.org. Questions? Email community@flok.org. ■

FINDING LOW PROTEIN FOODS AT GROCERY STORES OR ONLINE

Sandy Van Calcar, PhD, RD
Dietitian, Metabolic Genetics Clinic
Oregon Health & Science University

There are many foods at grocery stores that are low enough in protein to include in the diet for MSUD. Some of these foods can also be purchased online. Although these foods typically aren't as low in protein as products available from specialty low protein companies such as Cambrooke Foods or Dietary Specialties, they may be more economical to purchase. Some lower protein food ideas can be downloaded from https://msud-supportorg.stage.site/wp-content/uploads/2023/09/MSUD-Low-Protein-Foods-Handout.pdf.

Breads, pastas and other grains.
Looking for foods labeled as "gluten-free" can be helpful to find lower protein grain products. Gluten is a protein that is naturally found in wheat, barley and rye. Any food made from these grains will likely be higher in protein than gluten-free products. Baked products that use rice or oat flour are often lower in protein than those made with wheat. Gluten-free pastas are also often low in protein. As always, it's important to check the "Nutrition Facts" on the package for protein content since other higher protein ingredients may be used in these foods, even if they are labeled as gluten-free.

Cereals
Some ready-to-eat cereals are lower in protein than others. Look for those that are made from rice or oats as these may be lower than cereals made from other grains. There are gluten-free cereals that may be available with other gluten-free products in the grocery store.

Dairy/Milk substitutes
Non-dairy, plant-based dairy substitutes are easy to find in grocery stores, usually next to cow's milk and milk-based products. In general, any dairy substitute made from plants will be lower in protein than milk-based products. Rice, oat, almond, coconut, and hemp milks are much lower in protein than cow's or other animal milks. These milks typically contain about 1 g of protein (about 80 mg leucine) in 1 cup compared to 8 g of protein (760 mg leucine) in 1 cup of cow's milk. The one exception to this is soy milk, which contains just as much protein as cow's milk.

Products such as cheese, yogurt, ice cream or cottage cheese can be made from a lower protein milk substitute and are also becoming easier to find in grocery stores. Most brands of milk substitutes are fortified with calcium so they contain a similar amount of calcium as from cow's milk. However, compared to cow's milk, most plant-based substitutes are not fortified with vitamin D. Especially if you do not take a medical formula that contains vitamins and minerals, it is a good idea to discuss your vitamin D intake with your metabolic team.

Desserts
Substitutes are available for desserts containing higher protein ingredients. These products are often made with plant-based milk substitutes to replace ice cream, include carob or other alternatives to milk chocolate and decrease the amount of nuts that are added. Baked desserts can be made using lower protein grain alternatives as well.

Many of these products are made by smaller companies and are often only available in certain parts of the country. Talk with your metabolic dietitian to get other ideas for these foods that are specifically available in your area. ■

BEST DIETARY PRACTICES FOR DIET POST LIVER TRANSPLANTS

Elizabeth King, MS, RD, LDN, CNSC, Transplant Dietitian, UPMC Children's Hospital of Pittsburgh
Sharon Strohm MBA, RD, LDN, Clinical Dietitian, UPMC Children's Hospital of Pittsburgh

There is finally freedom from MSUD diet restrictions after a liver transplant.

Nutrition therapy goals post-transplant include healing, attaining a healthy height and weight, and incorporating a wide variety of healthy foods into the diet. These goals can be challenging for patients as their diet shifts from the highly

restrictive MSUD diet to a regular, unrestricted diet in which they are able to eat foods that were previously forbidden.

After transplant, it is important to consume adequate protein for post-surgical healing. The traditional MSUD diet is heavy in carbohydrates and inadequate in high quality protein, therefore patients must rely on an oral MSUD formula for most of their nutrition intake. It is difficult for patients of all ages to transition to consuming quality protein sources such as beef, chicken, pork, turkey, tofu, soy milk, lentils, eggs, milk, yogurt and cheese due to their taste and texture. Patients will often accept a milk, soy or pea-based nutrition supplement to meet their protein requirements until they are able to incorporate protein foods into their daily routine.

Oral aversions may add to the inability to consume adequate food. It is important to enroll in an early intervention program or occupational therapy program prior to a transplant depending on age. If your child has aged-out of these programs, it is imperative to receive outpatient oral therapy sessions. After transplant, these programs may be resumed. Age-appropriate formula and nutritional supplements will be utilized to assure adequate nutrition post-transplant. Standard infant formula for infants or oral commercial nutrition supplements (Carnation Instant Breakfast) are good to use while the patient is increasing variety in diet.

Tube fed patients will be transitioned to an age-appropriate formula. It is recommended that these patients continue to receive oral therapy as they gradually transition from a tube feeding to an oral diet.

An unrestricted diet is allowed after liver transplant for MSUD. Including a wide variety of protein foods into the diet is essential to normal growth and development. This presents challenges to both the patient and caregivers. A supportive transplant care team can support these challenges through education and intervention when needed. ■

PETUNIA: GENE THERAPY IN AN MSUD COW

By Heather Gray-Edwards, DVM, PhD
University of Massachusetts Chan Medical School

"Petunia", a calf with MSUD, was treated with human sized doses of AAV gene therapy when she was 2 days old, approximately $1M dollars' worth of AAV gene therapy. This life saving virus was made possible by the generosity of Dr. Guangping Gao and the hard work and dedication of Dan Wang, Jiaming Wang and the UMass Chan Vector Core.

Like children with MSUD, Petunia was born normal but became very sick quickly after birth due to the high amounts of protein and branched chain amino acids in the colostrum. She required around the clock care provided by the Gray-Edwards lab Veterinary Team and Clinic for Special Children (CSC) clinicians. Once stabilized, "Petunia" was fed the same MSUD diet adult patients with MSUD are fed and gradually transitioned to a normal diet.

Petunia is now 2 years old and is a normal heifer calf. She lives on a pasture at the Cummings Farm at Tufts Veterinary School. Petunia's biggest problems are benign excessive tear production and arthritis that is managed with joint injections. Overall "Petunia" has taught us that AAV gene therapy is as effective as liver transplant for MSUD up to two years after gene transfer. Therefore, this therapy offers an alternative to liver transplant.

Although the systemic delivery of AAV gene therapy for MSUD may be effective in treating peripheral disease in the body, magnetic resonance (MR) studies have shown that it may not be treating the brain. "Petunia" has the same MR spectroscopic abnormalities as MSUD patients after liver transplant (low glutamate levels). These data suggest that the gene therapy strategy used for Petunia may not be sufficient to treat the neurocognitive conditions experienced by MSUD patients on formula or after liver transplant. To further refine the treatment, we are pursuing higher doses and alternative delivery routes to better treat the MSUD brain. Dr. Gray-Edwards has submitted a grant to the National Institutes of Health to support this work.

To develop a better MSUD therapy we are planning to treat more calves with MSUD. We have run into a few challenges along the way. We have lost two MSUD calves due to failure of passive transfer, where plasma transfusion was insufficient to replace the antibodies provided by colostrum. These MSUD animals died of pneumonia and clostridium infection less than 24 hours after birth. We also had a high embryo mortality rate after embryo biopsy, which is necessary to determine MSUD status. To address this issue, we have started regularly implanting embryos without biopsy at a local dairy farm. We have three pregnancies with due dates in Fall, 2023 and Spring, 2024. The MSUD status of these calves is unknown. We have also acquired two new MSUD carrier cows, who are regularly producing embryos for this project. These new carriers are mini-Herford breed, thereby reducing the need of AAV by half. This allows the project to treat more animals to better understand the effects in patients.

Overall, UMass Chan and the CSC are committed to developing an AAV gene therapy for patients. Your continued support of this project is greatly appreciated! ■

PROGRAM STATUS UPDATE FOR CDX-6210, AN ORALLY DELIVERED ENZYME THERAPY FOR THE POTENTIAL TREATMENT OF MSUD

Codexis, Inc., Redwood City, CA
By: Kristen Skvorak, PhD

Codexis, a leading enzyme engineering company, recently described the discovery and validation of an orally delivered leucine decarboxylase for the potential treatment of MSUD.

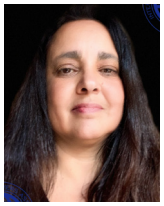
You can read the full report, published in the Journal of Inherited Metabolic Disease in July of 2023, here: https://onlinelibrary.wiley.com/doi/10.1002/jimd.12662. A complementary summary of Codexis' approach to and goals for this program can also be found in the MSUD Family Support Group Fall 2022 Newsletter.

Our modified, or as we say at Codexis - "evolved", leucine decarboxylase enzyme, also known as CDX-6210, was designed with the goal of surviving the harsh, acidic environment of the stomach and function in the small intestine to break down leucine, either coming from diet or enterorecirculation from the bloodstream. In healthy monkeys and MSUD mice, when CDX-6210 was given with a high-protein meal, a decrease in plasma leucine and leucine's associated branched chain ketoacid, KIC, was seen compared to animals only provided the high protein challenge. Importantly, CDX-6210 reduced leucine and KIC in the brains of MSUD mice, which is the site of leucine toxicity in this disorder. Excitingly, CDX-6210 also demonstrated that daily administration with a protein meal could stabilize and maintain blood leucine levels at pre-challenge baseline in the MSUD mouse model while blood levels of protein-challenged animals significantly elevated. Collectively, these data support further exploration of this enzyme as a potential therapeutic option for treating MSUD.

In July, 2023, Codexis announced its intention to streamline operations, focus on its core enzyme engineering capabilities, and to discontinue investment in its biotherapeutics programs. Due to this corporate decision, Codexis is actively seeking opportunities to partner this program in order to allow for its continued development. Initial preclinical and CMC work has been completed for CDX-6210, and the compound is now ready for material scale-up and preparation for IND-enabling toxicity studies, which are required to show test safety prior to the initiation of a first-in-human clinical trial.
If you are interested in learning more about CDX-6210 please initiate contact at www.Codexis.com. ■

DEVELOPMENT OF A NON-INVASIVE, PORTABLE BCAA MONITOR

By Dr. Lital Alfonta and Dr. Orna Staretz Chacham
Ben-Gurion University of the Negev and Soroka Medical Center, Beer-Sheva, Israel



We aim to develop a reliable biosensor for monitoring the branched chain amino acids (BCAAs) leucine, isoleucine, and valine to aid in the treatment of MSUD. Electrochemical biosensors have the advantage of being sensitive, fast, and requiring minimal sample volumes, making them suitable for point-of-care testing and non-invasive monitoring.

Currently, monitoring amino acid levels requires blood sampling and analysis in specialized laboratories. This process can be time-consuming and inconvenient for patients, particularly when frequent monitoring is needed such as during times of illness. Our goal is to develop an electrochemical biosensor which could potentially be used for non-invasive BCAA monitoring using saliva, which is easy to collect. The test could be performed by caregivers or even patients themselves, providing quick and accessible results for timely treatment decisions similar to a glucose monitor. While currently in the early stages of development, this biosensor will utilize amino acid-specific enzymes to ensure specificity and efficient electron transfer, improving sensitivity for detecting amino acid concentrations over a wide range from low to high.

The ultimate goal of this project is to improve patients' quality of life and reduce hospital visits and admissions by enabling easy and frequent monitoring of amino acid levels in bodily fluids, preferentially in a non-invasive manner.

Editor's Note
This Portable BCAA Monitor is supported in part by a grant from the Million Dollar Bike Ride. ■

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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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