MSUD
FAMILY SUPPORT GROUP

WWW.MSUD-SUPPORT.ORG

MSUD NEWSLETTER

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FORMULA UPDATES

Nutricia North America is pleased to confirm that all MSUD formulas are in stock and available. They are phasing out Complex MSD Junior and are working with healthcare professionals to transition children to either MSUD Anamix Early Years or Complex MSD Essential. They ask that you please contact your metabolic healthcare team if you have any questions regarding which product would be appropriate for your child. Call Nutricia at 1-800-365-7354 or email NutritionServices@nutricia.com if you have any questions about their MSUD products.

Abbott reports that new batches of Ketonex I and Ketonex II will be ready to ship in April, 2023.
Vitaflo reports that MSUD Gel, MSUD Cooler, and MSUD Express Plus are all available.
Cambrooke reports that their MSUD Vilactin AA Plus Powder 15 is available. ■

THE MSUD FAMILY SUPPORT GROUP SPORTS A NEW LOGO

The MSUD Family Support Group is made up of children and adults with MSUD traditional and non-traditional families.

Graphic designer Krista Trusz has redesigned our logo in a way that better represents our families and our mission. The new logo is somewhat abstract as compared to our previous one. It represents the human connection to the disease on a global level represented by a half circle shape of people providing support to each other. We hope that you like it as much as we do.

MSUD Family Support Group Board of Directors ■

MSUD REGISTRY UPDATE

By Karen Dolins, Research Lead

Thank you to all who took the time to complete our MSUD Registry with Sanford CoRDS! 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 importance of this registry to all with MSUD cannot be overstated.

When reviewing your responses, we noticed that some participants skipped several questions, possibly because they were unsure of how to answer them. Our questionnaire has now been updated to make it easier to understand. 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@sanfordcords.org to update your information. The MSUD Registry will be most effective if it is updated annually.

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. We are in the process of translating our questionnaire into Arabic and will let you know when this is available.

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 ■

Inside This Issue:

Sections

FROM THE PRESIDENT'S DESK

By Sandy Bulcher

After a long overdue phone conversation with a fellow MSUD mom that I've known for years, I've been thinking about how important it is to support each other in our MSUD journey, even if our journeys look different. We can understand and empathize with each other's challenges, often more than family and close friends.

Supporting each other involves interacting and connecting. Many of us do not live near another MSUD family which makes building relationships with other families challenging. Thankfully social media makes it possible for us to meet, share our story, exchange information and develop relationships online. We all know, though, that social media has its limitations and often doesn't replace face to face and heart to heart interactions.

Unfortunately, it's been 5 years since we had our last in person symposium and I'm concerned that some people have not been able to connect and receive support, which is especially important for our new families and MSUD teens and adults. There is good news, however! We are planning an in person symposium most likely in the Washington D.C .or Baltimore area, in late June 2024. Symposiums play a vital role in connecting and engaging our community and I would encourage you to plan to attend. There will be more symposium details to follow in the upcoming newsletters and on our website www.msud-support.org.

In the interim, there are a number of ways to get information and connect with other families. Information is available on our website and our official MSUD Facebook page. You can find this by typing "MSUD" in Facebook search and clicking on "MSUD Family Support Group" under "Pages". We also periodically send out e-blasts and you can sign up to receive those on our website. The MSUD board has hosted several virtual sessions already and we plan to continue those. Watch for details about the next session.

To get or stay connected, we have a private interactive Facebook group. It can be found by typing "MSUD" in the Facebook search and clicking here MSUD Support Community (not the official MSUD Family Support Group) | Facebook.click here . You will need to request to join this group. If you are interested in connecting with MSUD transplant families, you can join the "MSUD Transplants" in the Facebook search and request to join that private group. Another form of connecting is through virtual meetings. Susan Needleman, an MSUD adult, hosts virtual meet-ups. She hosts one independently for teens and adults living with MSUD. She also runs one under the direction of the New England Connection for PKU and Allied Disorders for parents and other caregivers of those with MSUD and similar disorders. If you would like to participate in the teens and adults one, email Susan at susanneedleman.msud@gmail.com. If you would like to attend the caregiver one please email her at necpad.org@gmail.com. Thanks, Susan, for all that you do for the MSUD community!

If you are not currently a member of our support group and included in our database, please call me (740-972-5619) or email me (sandybulcher@gmail.com) with the following information: Parent(s) please include your name, address, email, phone number and MSUD child(s) name, date of birth and type of MSUD. For those with MSUD, please include your name, address, email, phone number, date of birth, and type of MSUD. It's important that we have an accurate database for education, support and research purposes.

I look forward to seeing you at the next symposium and as always, please feel free to call or email anytime.

Sandy Bulcher, President MSUD Family Support Group and mom to Jordan, age 33, Classic MSUD ■

FROM THE EDITOR'S COMPUTER

By Susan Needleman

As I sit down to write this, it is Friday, March 24, 2023, exactly three years since the "COVID-19 Lockdown" started for many of us in the United States. I knew this day was coming and have been reflecting a lot on everything that has happened and how the world has changed. During these same three years many have had graduations, started new jobs, found a new formula they liked, found new recipes and favorite foods, joined virtual meet-ups and connected with others who have MSUD. There are very few, who can say the last 3 years were easy. What ever happened in your life in the last 3 years, you have persevered. You found a way to find and start your new normal. Hopefully, you found a way to go back to how things were before COVID-19, or found a new substitute if you have not. In the MSUD Community we are stronger than ever, the MSUD Facebook pages are as active and alive. Studies are being done to improve all of our lives, and the MSUD virtual meet-ups are meeting monthly. There are plans for a symposium next year and finally, MSUD formula is coming back in stock! We are living our lives and hopefully things are better for you then they were three years ago today. I am excited for what the next three years will bring to our community! ■

FROM EDITOR'S ASSISTANT'S WRITING NOOK

By Nikolai Rudd

Hello everyone! I'm writing a little over three years after the start of the pandemic. I've known many people who have gotten COVID-19. Some have died, while others seem to have brushed it off quite easily. And I see them doing things that I haven't been able to do since this pandemic began. But, what do I do "IF" I were to get COVID-19, being an MSUD Transplanted person with a compromised immune system??

My parents got COVID-19 while on their first trip abroad since the start of the pandemic. This prompted me to contact my Transplant Coordinator and Doctor to ask "if I ended up coming down with it", would I be able to take Paxlovid (like my parents) or one of the other oral medications for it? It was quite fortunate that I called, because it turns out the answer was a bit more complicated than I had thought.

You see, since I was vaccinated, boosted, and had taken EVUSHELD (which gave six months of protection), I was planning on testing my comfortability with things and trying to get back to a more normalized social and hopefully work life. My Transplant Coordinator told me that Dr. Chopra, (my Transplant Doctor at UPMC), said that because of my compromised immune system and the medications I was taking, I couldn't take any of the oral medications for COVID-19 and could only get the Infusion Therapy. However, they conveyed to me that the insurance companies stopped covering the cost of the Infusion Therapy, since the companies weren't convinced how much it helped. This policy would leave me to pay for the cost of the treatment out of my own pocket, costing roughly $4,000. My jaw dropped wide open!!

Not only was there no affordable or covered medication for this virus, but I couldn't even AFFORD to get COVID-19 because of the very cost of the ONLY treatment I was allowed to take.

There goes my attempt at getting back to a more normalized life, and having the chance to "actually" hang out with my friends from the dance studio (though outside of the studio itself). I have had to turn down going to clubs, dinners, or just doing things together with them on the weekends. My Transplant Doctor said they couldn't "tell" me to keep living as if the pandemic were still at its highest threat, but he did say I should minimize my "risks". As a result, I had to protect myself by reverting back to keeping up this hermit-like lifestyle . . . which is very lonesome. The only face-to-face interaction I have with people is for 1-2 hours of dancing classes, at the studio, since I live alone. This tends to make me wonder how I can actually live a fulfilling life without getting bogged down in depression and anxiety, until there is a safe, affordable, alternative to protect those of us with a compromised immune system. If any other MSUD Transplanted people feel you would like to contact me for support and to share ideas, you are welcomed to, my contact info is on the last page of the newsletter. ■

ADVOCACY UPDATE

By Jordann Coleman

2023 brings a couple pieces of legislation that are important to the MSUD and rare disease communities. The Medical Nutrition Equity Act (MNEA) ensures public and private insurance coverage for medically necessary foods when prescribed by a physician. The bill is of vital importance to the MSUD community. It was initially introduced in 2018 and was re-introduced to the 117th Session of Congress in 2021. The prior version of the bill gained a high level of support from many members of Congress. The newest version of the bill will be reintroduced later this Spring to the 118th Session of Congress.

We need your support to move this bill forward. We need more MSUD stories to show how essential this bill is to our community. Go to https://nutritionequity.org/share-your-story/ to share why having coverage for medical nutrition is important to you and your family. Personal stories are critical in gaining legislative support. Stay tuned for additional calls to action once the bill has been reintroduced.

In addition to pursuing the legislative process, The MSUD Family Support Group was one of 33 organizations who endorsed a request to include medical nutrition as an Essential Health Benefit (EHB) with the Centers for Medicare & Medicaid Services (CMS). By deeming Medical Nutrition an EHB it could better address the gaps in coverage between insurance plans and individual states and help with affordability. The Biden-Harris Administration has also acknowledged the essential nature of formulas and other nutritional supplements for children and adults with metabolic disorders in their "National Strategy on Hunger, Nutrition and Health". As CMS reviews the EHB, the ask is for it to take aim at the systemic barrier of access and affordability of medically necessary nutrition for those with metabolic disorders.

Lastly, the Newborn Screening Saves Lives Reauthorization Act passed in the House last summer, but did not get through the Senate before the prior legislative session ended. The Act reauthorizes existing federal programs that assist states in improving and expanding programs, supports parent and provider education, ensures laboratory quality and effective surveillance, and facilitates adding of conditions to the Recommended Uniform Screening Panel (RUSP). Look for calls to support this bill when it is reintroduced later this year. ■

EMORY UNIVERSITY'S METABOLIC CAMP

By Kemora Foster, 15 Years Old, Classic MSUD

In June 2022, I went to Emory University's Metabolic Camp. There I and the other campers were put into groups where we talked about what we ate and kept track of it every day to make sure we had the right amount of protein. Some campers were told by their doctors to only eat 2-6 grams of protein a day because the lower levels they consume, the lower leucine levels they have, and the healthier they are. While some could have a little more than that, everyone was very restricted. We also learned how to count protein, and what happens to people with MSUD when they have too much protein. Some people may feel and act crazy, think and do bizarre things that they can't control, feel sick and weak.

We also had information sessions about different topics related to MSUD. One was about liver transplants. I learned a lot there. I learned that some undergo a liver transplant because they don't want to drink formula anymore and some just don't want to deal with MSUD. When they have a liver transplant, some people can donate their liver to help someone else. But either way, the MSUD patient getting the transplant receives a new liver into their body.

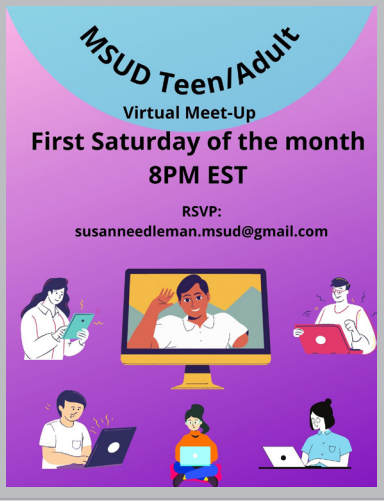
I would highly recommend Emory University's Metabolic Camp. Anyone who goes will find people that have MSUD and PKU there. They will find it easy to get along with everyone, because they have so much in common with them, and they won't feel alone anymore. ■

VIRTUAL MEET-UPS

By Susan Needleman

Caregivers/Parents: In addition to editing the MSUD Newsletter, I am also the Vice President of the New England Connection for PKU and Allied Disorders. This Spring, under their direction, I will begin to Co-Chair Caregiver/Parent quarterly meet-ups starting Monday, May 22, at 8PM EST. To find out more information and to join please email: necpad.org@gmail.com

Teens/Adults: Want to talk with others that have MSUD? Teens and adults with MSUD are invited to join us the first Saturday night of each month at 8pm EST/ 7pm CST on Zoom. At these meetings you will be welcomed by a friendly group of others like you, with MSUD. If you would like to attend please email me at susanneedleman.msud@gmail.com.



ADVOCATING FOR MSUD ON RARE DISEASE DAY

By Hannah Dolins, Classic MSUD Age 28

My doctor recommended me as a representative of an adult with MSUD for the Rare Disease Day Panel at Albert Einstein Medical Center entitled "Adults with Rare Disease as Advocates". I was one of four women that presented with their doctors. Of the other three, one had Gaucher Disease, one had Williams Syndrome, and one had Late Onset Tay-Sachs Disease (LOTS). I was up first. My doctor, Dr. Melissa Wasserstein, and I were introduced and, since we didn't have slides and preferred to just talk, we sat at the little table on the stage and tag-teamed about everything MSUD: The metabolic defect, what causes our levels to go up, how it feels when we're sick, psychological issues, traveling, limitations, etc. We told the audience how I advocate for MSUD by teaching doctors, medical students, residents, dietitians, and genetic counselors about MSUD and what it's like from the patient's perspective in the hopes that it sticks with them.

The presentation happened the day after I had a very stressful return from the Cayman Islands. I relayed what happend to the audience:

On February 24th 2023, I went to the Cayman Islands for the first time for a good friends' wedding. On February 27th, I tried to go back home. Boy was that difficult. I went on the security line with my metabolic formula as usual, let the agents know it was medical formula, and then I was stopped. I was not allowed through security with my formula because it did not fit into their 3 ounce bags. I told the person in charge that it was a metabolic formula and showed them the letter from my doctor explaining why I need to travel with it. I told them that I needed to take it with me, that if I didn't have it I would get very sick and could die. They said I didn't have the right documentation to take my formula through. They needed the exact prescription. I had to call Verizon and use them as a switchboard to try to get in touch with my doctor, who didn't answer (she was in clinic) and finally used them to get in touch with my mother. My mother tried to talk to the head security guard but he refused to talk to her. She finally told me that she'll try to get in touch with Melissa, but if it got too close to boarding time, I should just dump the formula out and board. That's what I ended up having to do, throwing my life-saving formula in the trash. After I got on the plane, Melissa called and asked if I had any powder, to which I replied I did not, as I hadn't expected to be away longer than a couple days. I prefer to take my formula with me already made when traveling. Meanwhile, unbeknownst to me, Melissa was conferring with Dr. Diaz from Mt. Sinai to see who they could contact in Miami in case emergency measures were needed when I landed (I had a layover in Miami). Thankfully, they weren't. When I landed in Miami, I confirmed my current formula measurements with my mother so she could have it ready for me when I landed, which she thankfully did, and was able to catch a slightly earlier plane home. The first thing I did was have formula as I hadn't had any in over 12 hours.

In addition to families of people with rare diseases, there were doctors, medical students, and other clinicians in the audience. After my presentation I was told that my words were very powerful and that they weren't likely to forget it. My main point was, always listen to your metabolic patients as they will know their disease and how it affects them better than you will. My other recommendation was that they should not be afraid to admit they know little/nothing about a disease; your patients can help you. ■

CANADA

Do you live in Canada? Did you know there was a support group just for families there? The Canada PKU + group recently expanded to include MSUD! This group provides their members with MSUD updates, resources, and offers networking and support opportunities. They now have a designated MSUD Chair to help you with everything along the way!

To find out more visit canpku.org.

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https://www.linkedin.com/company/canpku/

Want to meet us? Come to one of our events!
We offer bi-monthly virtual meet ups for adults and another one for caregivers! Sign up go to https://www.canpku.org/Events

Or Join Us In-person
June 2-4 Camp MagniPHEque, Redberry, SK
www.canpku.org/magniPHEque
Sept 16- Ottawa, Ontario
www.canpku.org/OntarioRegional
Sept 30 - Riverview, NB
www.canpku.org/AtlanticRegional
Travel Scholarships available
https://www.canpku.org/Travel-Scholarship ■

PORTER GOES TO GERMANY

By : Emily Anderson
Mother to Porter, 18 Years Old, Classic MSUD

Jameson and Porter

Last spring, Porter spent five weeks in Wildeshausen, Germany with his high school class. It was just about glitch-free, and it was a life-changing experience for him! Here's how we did it.

Porter Roth was diagnosed with Classic MSUD when he was three days old. Since then he has been drinking "juices" (as we call them) and eating a low-pro diet. He's had a few scares and plenty of visits to the hospital over the years, but he lives an otherwise normal life and is now in charge of managing his own diet. He can consume 12 grams of natural protein a day.

When he got the opportunity to go to Germany on a class trip, I knew we had to say yes. Not only was it important to me that he see more of the world, but he needed to know that he could lead a rich and exciting life.

To prepare, I sent the teacher in charge tons of information about MSUD and all of Porter's needs. The teacher then found a host family for Porter that was willing to take it on. With the help of our clinic, I tracked down a specialist in inborn errors of metabolism who worked only an hour from Wildeshausen. She agreed to be on call for Porter, and she was very receptive and supportive. Honestly, the hardest thing about preparing was getting copies of his travel and emergency documents in both English and German. At some point, we did have to find a translator.

Before Porter left, I shipped a case of his juices to his host family. He then checked an extra suitcase with low-pro food and more juices, and he had a carry-on bag with enough of everything for two days. While the students traveled, they made sure to eat at places with potatoes, fruits, and vegetables that Porter could have. As soon as he arrived, his host family took him to a grocery store where he picked out all of the low-pro food he would need.

Going to Germany was the best experience Porter has ever had. The town was quite small and his host family included a father with two sons. The father and older son spoke fairly good English. All of them were kind, generous, and funny. He went to school with his host brother during the week. The classes were all in German with the exception of the English language class, in which Porter and his friends were the experts!

On the weekends, Porter and his class traveled to other cities. In Vienna, Austria Porter visited the Hapsburg Royal Palace, saw many churches, and went on a tour of the city. He learned to navigate the Viennese metro system, and he even swam across the Danube river! The highlight in Vienna was buying three pounds of olives for only five Euros. He ate them right up.

His favorite trip was to Salzburg, Austria. The town was so old and beautiful and full of mountains. The students went to a museum and saw an old fortress turned into a monastery. The best part was eating dinner outside with his friends, next to a river, at sunset. In Berlin, Porter saw the German Parliament, lots of embassies, and the Berlin wall. He toured several other cities as well.

The worst trip was to Hamburg, but that was his fault. Porter and his friends were feeding geese near the bank of a river. He wondered what would happen if he tried to put food in the goose's mouth himself, instead of waiting for the goose to eat out of his hand. He found out what would happen when the goose bit his finger pretty hard.

Porter did catch Covid from another student while he was in Hamburg. The teacher called me immediately, and his host father came to get Porter that night. We all tracked his symptoms together, but they were very mild and he didn't need any intervention. It was certainly scary, but I was grateful that he had been boosted and that we had a specialist nearby just in case.

Throughout the trip, Porter was able to go to restaurants, where he ordered potatoes and vegetables. His teacher took him grocery shopping when they were on weekend trips, and Porter could usually find street food to eat. There were lots of olives and once even avocado sushi.

I'm so grateful that Porter was able to spend this time traveling. It broadened his views of the world, and it made him so much more confident about himself and how much he is capable of. He can't wait to go back, and I hope he does. ■

STRUGGLES OF AN MSUD ADULT

By Ryan Marvin, Age 22, Classic MSUD, Maryland, USA

I would like to take the time to talk about my struggles with Social Security Income (SSI) and the Department of Human Services (DHS) system. Since I turned 21, DHS was debating whether I was eligible for coverage of my MSUD formula, my SSI checks, and of course my healthcare. I spent the whole year on the phone with the government battling for my rights to have my formula covered by my insurance. There were obvious times where I was very close to running out of formula and facing severe health complications, including death as a result, but luckily it did not get to that point.

I had to take the distributor of my formula to court, which was very stressful for me. Stress, I was told, was one of the main factors of getting increased levels. I stressed out all year over this, which led to the aforementioned high levels, and of course other issues. These included being constantly drained and the mental damage it came with. My life was on the line, I could have died I spent most of my days either depressed, angry, or super worried and anxious to get this over with. I did not know where I was going to go with it. I did not know if I was going to run out of formula and die before I knew if I was going to win or not. I was worried that the government had the upper hand (which they always did).

It is the system we, as citizens, have to follow and play fair to. You just cannot say your piece and be on your way. I had to cooperate with their system and with that came a lot of patience, even though it was getting close to a time where I thought I was going to run out of formula. No one should have to face this just because they have turned 21 and have MSUD. The recent formula shortage put some people with MSUD in crisis, while some were already in crisis beforehand, struggling to afford their formula. MSUD affects everyone differently. Some of us face many challenges with the disabilities that MSUD can cause. These challenges and disabilities can limit the choices we have for work, and make it harder to find a job that pays at least $1,200 a month.

I have been able to tell when my levels are high by the smell of maple syrup, which is an indication that someone with MSUD needs medical attention. These past two years my levels have gone up more than they used to. My stress and dehydration contributed to these fluctuating higher levels. I was told that growing up was not going to be easy, especially when my levels get high. I was not informed that the smell of maple syrup was going to be such a strong presence. The smell has become a frequent occurrence. My ear wax, sweat, and urine can all smell like maple syrup when my levels are high. I have had to have my friends smell me for possible maple syrup in public because the smell is so common. Sometimes I think I smell it but I am not sure if it was me, a candle, food, a cologne, or something else.

It's definitely not an easy job keeping track of this disorder and keeping track of having to deal with SSI and fighting for your formula. I felt like it was hopeless. It came to a point where it was becoming inevitable that something was going to happen but I stayed patient and kept appealing. I kept appealing and I kept dealing with it. Finally, I received the judges answers to the court case. I won! I got complete coverage of my formula but my SSI checks and my medical foods were taken away. I used to get a food shipment every 3 months too, about $500 worth. With that and the money gone it force me to try to find other ways to take care of myself.

Getting a job to pay for this food is only made more difficult by the pandemic. Job hunting is not a walk in the park. You have to look for a job everywhere. You have to find a job that you meet the expectations for. I have to fit this search in between taking care of my health and finding foods in the grocery store that I can eat, which of course costs more than higher protein foods. This has led me to - make some of my low protein meals from scratch so I can have more to consume and feel normal. I am restricted though due to the costs of these foods but it allows me to have more options. These challenges that I have had to face have made me want to spread more awareness about MSUD and growing up with it. Thank you for listening to my story. ■

MY STORY WITH MSUD

By: Azucena Gonzalez, Mom to Isabella, 7 years old, San Luis Potosí, México

Isabella was born on December 1, 2015 with MSUD. Her older sister does not have it. When Isabella was born and took her first screening something was off but they did not want to alarm me. A second screen was done which also came out abnormal.

The doctor looked for a geneticist in the same state where we live, who performed another test and the doctor only gave us a presumed positive. Isabella was put on a special formula, but didn't advise us exactly how much. I decided to go to Mexico City, guided by Dr. Marisol Piña, at the National Institute of Pediatrics where the Laboratory of Inborn Errors of Metabolism is located. The doctors, a group of women very dedicated to their profession, obtained a quantification of branched chain amino acids there again and they gave the diagnosis that Isabella had MSUD.

At that time she was already 6 weeks old, but her doctors told me that her disease was a variant of the classic form. She never had a strong relapse, and was never in a hospital but she did have some reactions to vaccines, complaints, etc. Mexico City is a six hour drive from where we live. For 8 months we consulted with the doctors there every week in order to keep track of her. Now, little by little, they have spaced out the consultations but they are requiring us to send blood samples to monitor her amino acid levels.

Right now Isabella is in the first grade of primary school and has led a completely normal life, except for the special diet that she must have. Her life is like that of other of her classmates:. She goes to school, does homework, learns to read, and loves to dance, talk, and is learning what she should not eat.

We want to thank Dr. Marcela Vela, Dr. Leticia Belmont, and her specialist(s) and nutritionists. I would also like to thank Sara Guillen and Lizbeth LP for continuing to work hand in hand with our family.

Hopefully Abbott's facility will be up and running again as normal, since they are the company that manufactures and distributes Isabella's formula which is what keeps her alive. ■

Vitaflo™ is grateful to provide solutions for the nutrition management of rare diseases to this valued community. Today we have exciting news to share about joining a sustainability movement called B Corp™. We will continue to offer all of the same products, and behind the scenes we have already started to pay more attention to how we treat our workers, the recyclability of our packaging and giving back to our communities.

So if it's important to you to use products made by companies who treat their people and planet well, we've got you covered.

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Kind Regards,
Your **Vitaflo** USA Team

MILLION DOLLAR BIKE RIDE 2022

MSUD RESEARCH AWARDS

By Karen Dolins, Research Lead

I am pleased to announce that our efforts to spark interest in MSUD research have been successful. The Orphan Disease Center at the University of Pennsylvania, which runs the MDBR and administers the awards, received 9 proposals for MSUD research. These were reviewed by a committee which included several members of our MSUD Scientific Advisory Board. The following projects were selected:

Development of neuroprotective gene therapy for MSUD by Dr. John Counsell, University College London
Dr. Counsell and his colleagues aim to develop gene therapy for the brain with the goal of reducing the neurological manifestations of MSUD. They will provide brain-targeted gene therapy to MSUD mice and compare their outcomes with mice receiving liver-targeted gene therapy.

Development of a leucine monitor by Dr. Orna Staretz-Chacham, Soroka Medical Center and Dr. Lital Alfonta, Ben Gurion University, Israel
Dr. Staretz-Chacham, a metabolic physician, and Dr. Alfonta, a bio-engineer, have teamed up to develop a device able to monitor blood leucine levels without requiring a blood test. This device will use electrical currents, a technique they have successfully used to measure blood glucose levels.

The selected studies target our key goals: development of a leucine monitor and finding a cure for MSUD through gene therapy. ■

MSUD FAMILY SUPPORT GROUP EXHIBITS AT SOCIETY FOR INHERITED METABOLIC DISEASES (SIMD) ANNUAL MEETING

By Karen Dolins, Research Lead

SIMD is an organization dedicated to increasing knowledge of, and promoting research for inborn errors of metabolism, such as MSUD. The meeting brings together clinicians working in the field and researchers seeking ways to improve treatment. This meeting has been identified as an important one for us to attend by our Scientific Advisory Board as we aim to increase interest in MSUD research. I was pleased to be able to represent the MSUD Family Support Group (assisted by my husband) at their 2023 meeting in Salt Lake City, Utah.

Our booth attracted a good deal of interest. Physicians and dietitians stopped by to learn about how we can help them support their patients. Researchers stopped by to learn more about our research priorities.
Dr. Jessica Gold, recipient of our 2021 Million Dollar Bike Ride grant, gave a brief presentation describing her findings from her study of executive function in adults with MSUD. She was a winner of SIMD's 2023 Travel Award. See her article in this issue of the newsletter describing her work.

MSUD Scientific Advisory Board members were active participants at the meeting. Dr. Irini Manoli served as Program Committee Co-Chair and Dr. Lindsay Burrage served as a member of the committee. Dr. Andrea Gropman was also present.

Several other patient organizations were in attendance, including HCU Network America (homocystinuria), Organic Acidemia Association, Propionic Acidemia Foundation, National Urea Cycle Disorders Foundation, and the National PKU Alliance. I was able to network with representatives from these organizations and discuss potential collaborative projects.

All in all, this was a successful meeting for our organization. I even got to wake up every morning to the view of snow-covered mountains outside my hotel window! ■

UPDATES FOR NEUROCOGNITIVE OUTCOMES AND QUALITY OF LIFE IN ADULTS STUDY

By Dr. Jessica Gold, MD, PhD

We have successfully recruited our goal number of adults with MSUD for this study!

The 28 adults enrolled in this study have the following characteristics:

18 females (67%)
26% individuals from underrepresented backgrounds
average age 34 years

Most adults (85%) were diagnosed in infancy when they showed symptoms of MSUD. Four individuals were diagnosed before symptoms started due to known family history, and 1 person was diagnosed in his 30s with intermittent MSUD. About 1/3 of the adults in this study are married and 5 have children. The highest educational level achieved was split: 25% graduated high school, 25% completed some college, 25% have a college degree, and 25% obtained a masters or doctorate. Adults with MSUD report a similar quality of life compared to healthy volunteers.

We evaluated neurocognitive outcomes using 2 techniques: Adults completed questionnaires about their executive function (planning, working memory, attention, impulse control) and their adaptive function (skills of daily living). They also played a collection of games on their computer or smartphone that tested executive function. Overall, we detected deficits in executive function and adaptive function in adults with MSUD compared to healthy volunteers. Individuals in this study with better executive function skills were more likely to have a full-time job, live with a spouse, obtain higher levels of education, and manage their health independently.

Interventions that strengthen executive function may be beneficial for individuals with MSUD, especially during adolescence when executive function develops and teens prepare for greater independence.

In addition to the above assessments, we also conducted interviews with adults with MSUD and their caregivers. These interviews focused on the unique needs of the adult community and gaps in the healthcare transition process. Emerging themes include a need for greater counseling on education, vocational, and financial supports; symptom changes in adulthood, and concerns about lack of knowledgeable adult providers.

Thank you to everyone who has participated in this study! Your input has been invaluable. We look forward to sharing our final results soon. ■

ACER MSUD NEWSLETTER UPDATE MARCH 2023

Acer Therapeutics Inc, a pharmaceutical company focused on the development and commercialization of therapies for serious, rare and life-threatening diseases with significant unmet medical needs, continues to work toward evaluating a potential new therapy (a form of sodium phenylbutyrate) to treat Maple Syrup Urine Disease. Acer is actively engaged in discussions with the Food and Drug Administration (FDA) to come to an agreement on the design of a formal clinical trial.

The goal is to initiate the first phase of the clinical trial activity in the early part of 2024. Leading up to that time, Acer will be working to identify the key healthcare providers who treat MSUD patients and seek their feedback and support for the study. Acer will also be looking to identify potential patients with MSUD to participate in the study when and if the time comes to begin recruitment.

Along the way, Acer will continue to work closely with the MSUD Family Support Group to seek feedback from the community and ensure that the needs of MSUD patients are heard. Acer remains committed to supporting the MSUD community as well as other communities that suffer from various metabolic disorders and other unique rare diseases. ■

June 2 - 4, 2023
Redberry, Saskatchewan

CAMP MAGNIPHEQUE

International Attendees Welcome
Meals, activities and fun included
Under 18 requires an adult
All Ages welcome
MSUD Welcome

www.canpku.org/
MagniPHEque

CANADIAN PKU AND ALLIED DISORDERS INC.
PCU ET MALADIES APPARENTÉES CANADA INC.
Because Knowledge Leads to Better Health
Parce que la connaissance mène à une meilleure santé
www.CANPKU.org

https://www.youtube.com/watch?v=Wp403MIl-tA

Metabolic Camp

A Holistic Approach to the Management of PKU & MSUD

Metabolic Camp June 19-24, 2023

Join our 29th Annual Metabolic Camp for summer fun, learning, and tools for managing PKU and MSUD!
More info at **metcamp.net**.

Camp Director: Rani H. Singh, PhD, RDN, LD
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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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