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MSUD NEWSLETTER

Published by the MSUD Family Support Group ▪ Volume 40-1 ▪ Spring 2022

THE MSUD FAMILY SUPPORT GROUP WILL AGAIN RIDE IN THE MILLION DOLLAR BIKE RIDE

By Ed Fischler and Herb "Butch" Foster

The Penn Medicine Orphan Disease Center (ODC) will again host its annual Million Dollar Bike Ride and has invited the MSUD Family Support Group to participate in the event. After enduring two years of the pandemic, we are hopeful that this outdoor event will return to the streets of Philadelphia on Saturday, June 11th, 2022.

The Million Dollar Bike Ride raises money for rare disease research. This event helps us support our goal to improve treatments for MSUD patients and move toward a potential cure for MSUD.

This will be our fifth year participating in this event. The MSUD"Team Scott" has raised donations to support research for improved treatments and a potential cure for MSUD. Each year, our goal is to fund at least one research project with at least $30,000 or more of donations, matched by an additional $30,000 by the event sponsors.

'BIKE RIDE' Continued on page 3

JOINT STATEMENT ON METABOLIC FORMULA SUPPLY ISSUES

3/15/2022: As you are likely aware, the FDA has placed a manufacturing and shipping hold on all products produced at Abbott's Sturgis, MI. plant due to reports of bacterial infection in several infants who consumed their Similac, Alimentum, or EleCare powdered infant formulas. Abbott has voluntarily recalled these products. All of Abbott's metabolic formula products are produced in this plant and are therefore affected by the manufacturing and shipping hold. There have been no reports of bacterial infection in individuals using these products and no metabolic products have been recalled. It is not known when this hold will be lifted.

The undersigned organizations have consulted with metabolic dietitians from across the U.S. and Canada, and representatives of Genetic Metabolic Dietitians International (GMDI), the Southeast Regional Genetics Network (SERN), Abbott, Nutricia, and Vitaflo, among others.

While we hope that the situation at the Abbott plant will be resolved quickly, and that the FDA will release the hold and allow Abbott to resume manufacturing

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MSUD FAMILY SUPPORT GROUP NEWS

FROM THE PRESIDENTS' DESK

By Sandy Bulcher

I've been involved with the MSUD Family Support Group for many years in various roles. Currently I am serving as President. My life has been enriched by the MSUD families that I've met through the years. I realized recently, though, that many of our families are new and we have not had the opportunity to meet in person or virtually so I decided to take this opportunity to share a little about myself and my family.

My husband Dave and I live in Columbus, OH and are the parents of two wonderful grown sons. Our youngest son Jordan, who is 32, has Classic MSUD. He was diagnosed at 17 days of age when newborn screening for MSUD in Ohio was not available. Fortunately, he does not have any negative effects from the late diagnosis and is able to live independently. He works as a neurology Physician Assistant at Ohio State. Our older son, Tyler, and his wife live nearby too and they recently welcomed their first child. Being grandparents has been a joy for us!

I am an RN and work full time in a busy surgeon's office. While I enjoy my job, it can be challenging to keep up with all of my work and support group responsibilities. My hope of a better life for those with MSUD, however, keeps me motivated. My husband Dave, works as an engineer and is also involved in the MSUD Family Support Group. He has served as Treasurer for many years.

I was looking forward to seeing my old MSUD friends and making new friends at the 2022 MSUD Symposium but the meeting planned for August 4-6 in Lancaster, PA has been canceled unfortunately.

This decision was not arrived at lightly. The MSUD Family Support Group Board of Directors remains concerned about our ability to safely bring our community together. Out of concern for the health and well-being of all and after weighing our options, we decided that it is in the best interest of this community to cancel this in-person event.

At this time, we do not know when or where the next symposium will take place but will update you when that information is available.

In the interim, we encourage you to stay connected and continue to support each other as you have been doing during these difficult times. We have held several webinars over the past year including a cooking event and an information session regarding our Patient Registry. We will continue to plan events that can be attended from the comfort and safety of your homes. Just like you, we look forward to the future when life is "back to normal". ■

EDITOR'S NOTE

From Susan Needleman, Co-Editor

Welcome to the Spring 2022 newsletter! I am excited to write to you for the first time as one of your Co-Editors. As a quick refresh from our last newsletter in the Fall 2021, I am 31 years old, with classic MSUD, and live in Massachusetts. Like many of you have recently experienced, I, too, struggled in the past few months with having to switch MSUD formulas and doubt with supply chain issues. I am happy to report that I did find something that works for me and I am sure those going through the journey of switching now will, too. When I was very young, there were only 3 formulas on the market. I am happy to report, there are now 19 to choose from in America! This is something 25 years ago many of us wouldn't believe was possible!

Formula has not been the only development for MSUD during this time. New studies have brought more education about our disorder and more types of treatments. Here at the MSUD Family Support Group, we are happy we can provide this information to you, along with advocacy projects, fundraisers, and of course your family stories. ■

Amber Raye, Co-Editor

We are in another challenging chapter in history with Ukraine and Russia currently at war and some MSUD formulas shipping being put on hold, causing significant shortages to MSUD families across the globe. I want to take a moment and say that as a community we are sending positive thoughts your way. We continue our efforts to provide you with research updates, family stories, and resources. Remember that our community is here to provide guidance and a safe place to get answers to questions regarding MSUD during this trying time. With love from your co-editor Amber Raye, my father, my fiancé Chris, and three fur babies (Loki, Valkyrie, and Iris-Lullaby). ■

'RIDE' Continued from front cover

Last year, over a dozen riders in the United States and Canada supported MSUD research by riding in their neighborhoods or near their homes. Some of their stories are highlighted below.

You can participate in this year's event in the following ways:

- Register (with fundraising) to ride in the event in Philadelphia by logging into the MDBR website (**https://www.milliondollarbikeride.org/registration**)
- Register (with fundraising) to ride in the event in your neighborhood as if you were riding in Philadelphia at **https://www.milliondollarbikeride.org/registration**.
- Donate on **https://msud-support.org/2022-million-dollar-bike-ride/** by clicking the "donate" button on the bottom of the page. No matter how much or how little you are willing to donate, every dollar counts towards finding better treatments and a cure for MSUD.
- Encourage your friends and family to visit one of these web pages and donate by clicking one of the buttons on the bottom of the page.

MSUD research is not a single project, but rather a journey. You can make this year's MDBR campaign an even bigger success. To contribute to the MSUD Family Support Group Research Fund, go to **https://msud-support.org/2022-million-dollar-bike-ride/** and donate at the bottom of the page. Participating in the Million Dollar Bike Ride or making a donation directly to the MSUD Family Support Group research fund moves us toward that goal. ■

'SUPPLY' Continued from front cover

and shipping of metabolic products, we have concluded that the most prudent course of action is for all metabolic patients to work with their clinicians to transition to a new formula as soon as possible as the timeline cannot be predicted.

Please be patient with your clinical teams as they reach out to you to facilitate this transition. This situation has placed an enormous burden on staff on top of their regular responsibilities. Please also understand that some metabolic disorders put people at higher risk of decompensation, and hospitalization, than others. Clinical teams must prioritize those patients, but rest assured they are working hard to make sure all patients have sufficient supply of a formula that is palatable and meets their nutritional needs. Your dietitians understand the challenges involved in changing formula and are doing their best to provide alternatives which will be acceptable to each patient.

We have been in contact with the FDA to express our concerns. They are well aware that metabolic formulas are essential to the health of our community and have issued a statement specifically regarding the metabolic products.

We will stay on top of this situation and keep you informed as new information becomes available.

Abbott Products Affected by the FDA Hold

Glutarex-1, Glutarex-2, Cyclinex-1, Cyclinex-2, Hominex-1, Hominex-2, I-Valex-1, I-Valex-2, Ketonex-1, Ketonex-2, Phenex-1, Phenex-2, Phenex-2 Vanilla, Pro-Phree, Propimex-1, Propimex-2, ProViMin, Calcilo XD, Tyrex-1, Tyrex-2, Similac PM 60/40

National PKU News | The MSUD Family Support Group | HCU Network America | National PKU Alliance | Propionic Acidemia Foundation | Organic Acidemia Foundation | Iowa PKU Foundation | National Urea Cycle Disorders Foundation | IPAD | NECPAD | Georgia PKU Connect | NOTA - Network of Tyrosinemia Advocates | Louisiana Metabolic Disorders Coalition | TN PKU Foundation ■

MSUD FAMILY SUPPORT GROUP NEWS

ADVOCACY UPDATE

By Jordann Coleman

The Medical Nutrition Equity Act was introduced in the House (H.R. 3783) and Senate (S.2013) last spring. The Medical Nutrition Equity Act would ensure public and private insurance coverage for medically necessary foods when prescribed by a physician.

This bill is of vital importance to the MSUD community. Currently there are 71 co-sponsors in the Senate and 74 co-sponsors in the House. The more co-sponsors this bill has, the better chances this bill will have of passing.

Do you know what 90% of these co-sponsors have in common? They have heard from multiple constituents that this bill is important to them and/or heard from the same constituents multiple times. It shows that being a squeaky wheel makes a difference!

We need your support to move this bill forward. We need more MSUD stories to show

how necessary the bill is to our community. Go to **www.medicalnutritionequityfor.us** to share why having coverage for medical nutrition is important to you and your family. You can also go to **https://nutritionequity.org/contact-congress/** to send an auto-generated email to your members of Congress asking them to become an original co-sponsor of the bill. Social Media is a great (and public!) way to ask for your Representatives and Senators' support (or thank them if they're already a co-sponsor).

Please share with your friends, family & social networks and ask them to contact their Congressional members. Together we can make coverage for medical nutrition a reality.

In addition, The Newborn Screening Saves Lives Reauthorization Act passed the House this summer. It is now in the hands of the Senate. The Act reauthorizes existing federal programs that assist states in improving and expanding programs, support parent and provider education, ensure laboratory quality and effective surveillance, and facilitate adding of conditions to the Recommended Uniform Screening Panel (RUSP). You can also contact your Senators to ask them to support this bill. ■

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Spring 2022

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TASTE

TRIAL

SUPPLY

SCAN TO REQUEST A SAMPLE

MSUD Maxamum is a medical food for the dietary management of Maple Syrup Urine Disease (MSUD) and must be used under medical supervision.

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RESEARCH

MSUD FAMILY SUPPORT GROUP RESEARCH UPDATE

By Karen Dolins, Research Lead

Promoting research to improve the lives of those with MSUD is a key goal of our organization. We have identified the following priorities for our efforts:

- Gene therapy aimed at repairing the genetic defect which causes MSUD
- Patient registry to help guide research
- Understanding the neuropsychological impact of MSUD
- Developing a home monitor for measuring blood leucine levels

I am happy to report that we are making progress in all these areas.

Researchers at UPenn continue to explore muscle and liver-directed gene therapy on animal models. Gene therapy is being examined in a cow model of MSUD cow at UMass, and Moderna is performing preliminary studies to determine the feasibility of messenger RNA (mRNA) therapy for MSUD.

Our patient registry is up and running. WE NEED EVERYONE TO PARTICIPATE regardless of the country you live in and whether you have received a liver transplant! This is your opportunity to tell researchers about your experience living with MSUD. If you have not yet done so, please access the registry by going to: **https://msud-support.org/msud-patient-registry/**. You can email me at karen.dolins@gmail.com if you have any questions about how to enroll.

One important piece of information we hope to learn through the registry is where people with MSUD live. We have partnered with Acer Therapeutics to explore this further. We hope to get a better understanding of the incidence (how many people are born with MSUD every year) and the prevalence (how many people have MSUD), where these people live, and where they are treated.

Work on the neuropsychological impact of MSUD continues. Dr. Rebecca Ahrens-Nicklas is continuing her work on mice studying how MSUD affects the brain and Dr. Jessica Gold is conducting a study aimed at assessing the neurocognitive outcomes and quality of life in adults with MSUD. Both researchers received financial support for their work from us through funds raised with the Million Dollar Bike Ride and each has written an article for this newsletter describing their work. Thanks to all of you that participated in our fund-raising efforts!

A portable monitor has been developed to measure levels of phenylalanine in the blood of individuals with PKU. This device is now entering clinical trials. If successful, we hope that the same technology can be used to develop a home monitor for measuring leucine levels. This would truly revolutionize care for those with MSUD!

Our scientific advisory board meets twice a year. During our December meeting, we reviewed current projects and discussed other potential areas for research. Our scientific advisory board helps us decide which projects we should pursue and helps us understand the results of ongoing projects. We are grateful for their expertise. Together we are planning to hold an MSUD Science Summit in August. We hope to bring together key researchers for a day of discussing current and future research. I will report on this meeting in the next newsletter!

Please don't hesitate to contact me with any questions regarding MSUD research. ■

UPDATES ON STUDY ON NEUROCOGNITIVE OUTCOMES AND QUALITY OF LIFE FOR ADULTS WITH MSUD

By Dr. Jessica Gold

Our study at the Children's Hospital of Philadelphia is in progress with some exciting changes. In the next month, we will be introducing the Adaptive Cognitive Evaluation (ACE). Developed at the University of California, San Francisco, ACE is a collection of web-based games designed to test thinking skills, decision-making, attention, and working memory. We will use ACE along with our current set of online surveys on life skills and quality of life. This will be the first time ACE has ever been used for people with inherited metabolic disorders and we look forward to introducing this new tool in adults with MSUD. The best part is that all the testing can now be done at home.

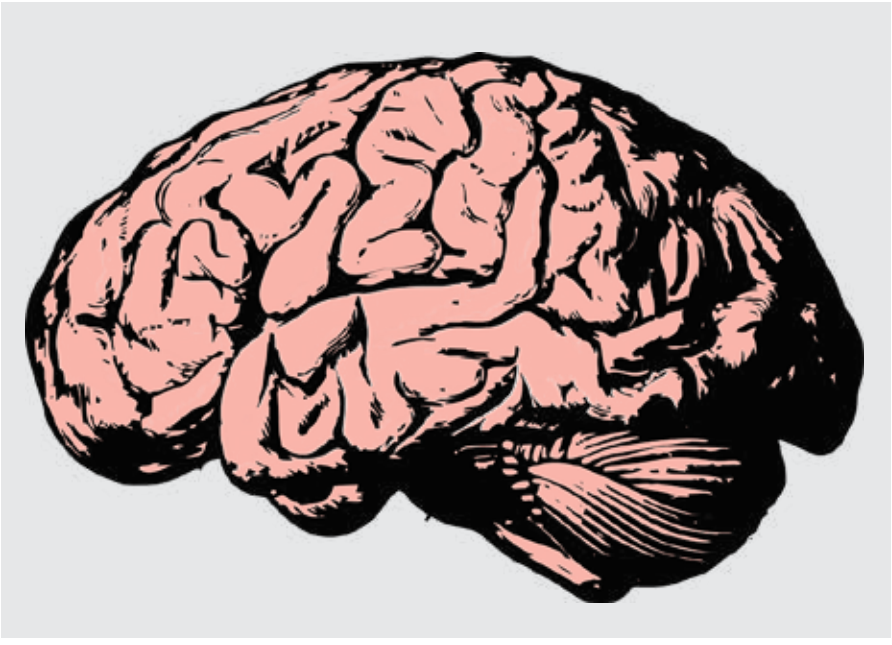
We are continuing to recruit for this study. Currently, 12 participants are enrolled. Overall, our goal is to enroll 25 adults with MSUD and 25 unaffected siblings or acquaintances. If you are over 21 years old and may be interested in participating in this study or have any questions, please contact Jessica Gold at goldj@chop.edu. ■

CAN WE IMPROVE MENTAL HEALTH IN PATIENTS WITH MAPLE SYRUP URINE DISEASE?

Jing Xu, PhD

What did the study look at?

Maple Syrup Urine Disease (MSUD) is a metabolic disorder of amino acids. Due to the inability to metabolize branched-chain amino acids (BCAAs) in MSUD, there is a build-up of these amino acids in the brain and a disruption of neurotransmitter production leading to psychiatric and neurocognitive issues.



These mental health issues may arise from brain development abnormalities in MSUD. To examine the early brain development in MSUD, researchers

looked at brains from healthy mice or mice with MSUD.

What are the key findings from this study?

Researchers found disrupted early brain development in the MSUD mice.

Why does it matter?

This finding will provide a new understanding of early brain development in MSUD. It will inform us of novel therapeutic strategies to tackle the mental health issues in patients with the disease.

Who conducted this study?

Dr. Rebecca Ahrens-Nicklas, Assistant Professor and Attending Physician at Children's Hospital of Philadelphia, led the team. Jing Xu, Amanda Kuhs, Jeet Singh, Sophie Siemsgluess, and Youseff Jakher assisted in conducting this study.

What were the results of this study?

The researchers made the following observations in the brains of MSUD mice:

- levels of BCAA were increased,
- levels of other amino acids were reduced,
- levels of major neurotransmitters were reduced,
- levels of several amino acid transporters were increased.

Amino acid forms proteins which are essential for early brain development. The researchers therefore looked at brain development.

- They found that MSUD mouse showed disrupted brain development.

What's next?

The team will explore whether drugs that increase acetyl-CoA levels can improve the disrupted brain development observed in an MSUD mouse model.

We will also be investigating brain function in a new brain-specific knockout mouse model of MSUD. We developed this model to reflect the experience of

well-controlled MSUD patients. Specifically, the mouse has normal blood BCAA levels but still has abnormal BCAA metabolism in the brain. Using this model, we will explore the mechanisms that contribute to chronic neurologic and psychiatric symptoms in MSUD.

MORE INFORMATION

Who funded this study?

- Penn Orphan Disease Center Million Dollar Bike Ride Grant https://www.orphandiseasecenter.med.upenn.edu/million-dollar-bike-ride
- MSUD Family Support Group https://msud-support.org/
- National Institute of Neurological Disorders and Stroke. https://www.ninds.nih.gov/

Where to learn more about Dr. Ahrens-Nicklas's work?

https://www.research.chop.edu/people/rebecca-c-ahrens-nicklas ■

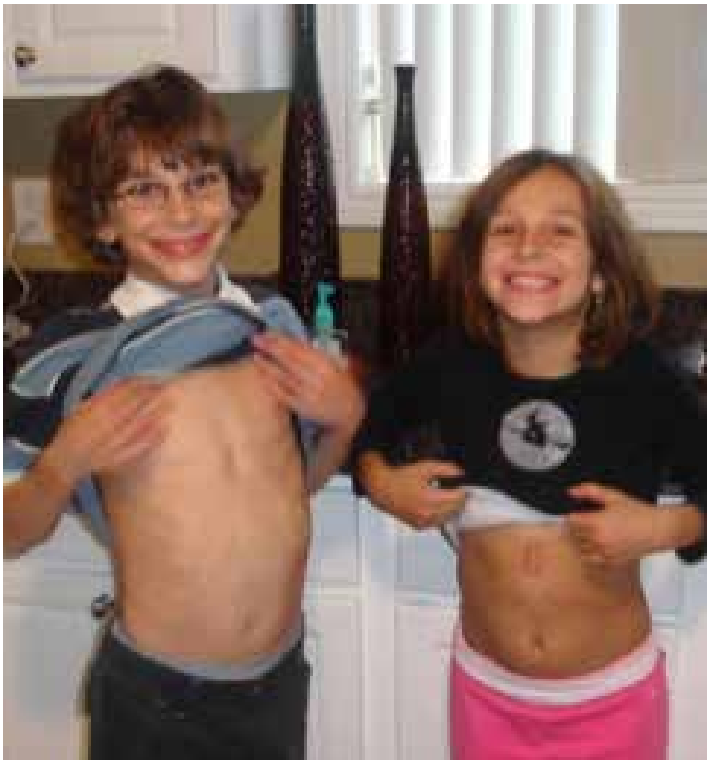
FAMILY NEWS

SYDNEY AND ZAC PINSKEY

AN MSUD LOVE STORY

By: Denise Pinskey, Mom, Zac, Age 26, Classic MSUD, Transplanted

Zac and Sydney



Zac and Sydney showing off their transplant scars as kids

***Syd*ney age 24: Classic MSUD, liver transplant recipient 8/10/2006,**

Zac, age 26: Classic MSUD, Liver Transplant recipient 6/27/2005.

Married on August 21, 2021.

The support group made them friends in 1998, Marriage made them family.

This support group is responsible for this life-long friendship and now the joining of families.

Sydney's Thoughts - I didn't think I'd ever have a job, or complete high school, or get married. And look at me now. One day I will drive like Zac. My transplant gave me a second chance at life. I remember coming to Zac's house to play in the pool, jump on the trampoline, and pet the bunnies. Now, Zac and I enjoy fishing and hunting at the Kelly cabin. I currently work at Kroger and enjoy helping out family members.

Zac's Thoughts – I remember Sydney and I always having fun together. We liked the same things, we have a lot in common: MSUD, transplant and the love of potatoes and ketchup! We also love going to the local fairs. I have always dreamed big and never gave up. Sydney and I are better together than apart. Work hard and good things happen. I am now working part time in oil reclamation and marriage is great. I enjoy helping coach high school wrestling

and football and spending time with my niece Julia.

Shawn, Sydney's Mom - There were fewer than 12 cases of MSUD in Michigan when Sydney was born. The hospital put me in touch with Zac's mom Denise. I remember receiving her letter telling me about her experience with MSUD and providing support and friendship. From that day on we've formed a friendship and kept the kids in touch whether it be visiting or preparing low protein meals. Sydney and Zac attended high school dances, family functions and were inseparable from middle school on. This marriage was the next step in their future.

Denise, Zac's Mom - I remember the first time we met Sydney and family. We were at our clinic visit at the University of Michigan, Mott's Children's Hospital. Sydney was a beautiful baby girl. After that, we mothers talked a lot and had many play dates. We would jokingly say that when they get older they could marry and we could take turns offering them support to live their best lives. At one gathering Sydney and Zac decided they wanted to marry. They were 6 and 8 at the time. We had to slow their time line down a bit. Sydney and Zac both won the most talkative/friendly award at transplant camp. They guided us in how life was going to go.

God definitely put these two together. Both families could not be any happier for them!

Thanks for reading our love story. We hope that our story will inspire you to dream big for your life and the lives of your children! ■

-Sydney and Zac Pinskey

KEEPING HOPEFUL THROUGH DARKNESS AND ISOLATION

By: Nikolai Rudd, Age 47, Classic MSUD, Transplanted

This past May, during our COVID pandemic and isolation, marked the Fifteenth Anniversary of my liver transplant, which turned my world around and upside down. It was 2006 when I got an early morning call to get to Pittsburgh for my "Domino" liver transplant. Since I did not have liver disease, my liver was able to go to someone else who was too low on the list and would have died before getting one if he had not received mine.

Prior to getting the call for my transplant, I had spent the first 32 years of my life living with Classic MSUD. Getting a transplant does not come without risks. My family and I had to stay in Pittsburgh for about a month before getting cleared to make the journey back home. One or two weeks after arriving home, I contracted Cytomegalovirus (CMV), which is an illness that most people get when they are young. It took me several months to bounce back. But the first real setback from the transplant occurred just after my three year anniversary. I woke

up one morning and could not stand up straight. I felt like I was 90 years old. Over the course of several months my primary and transplant doctors had me undergo a whole battery of tests over the course of many months. These included gastro-intestinal tests, MRIs, CTs, and several X-rays in Massachusetts where I lived and Pittsburgh. After ruling out many different possibilities, they came to the conclusion that my scar tissue adhesions were the culprit. Since there is no way to see scar tissue adhesions other than cutting you open again, it was a diagnosis through exclusion. And because my digestion had been slowed down a lot, they figured that the adhesions had started interfering with other organ functions. Though it was an educated guess, my transplant team figured that my adhesions were wrapped around part of my intestines.

Since I was already prone to getting these thick and ridge scar tissue adhesions, my transplant team warned that having another surgery to cut them was not an option, as it was more likely I would just get new ones in possibly worse areas. Despite this roadblock, my mother began to do extensive research to find non-surgical options. Eventually, she found a place in Gainesville, Florida, called "Clear Passage". They had developed a new deep tissue massage therapy that was said to assist in permanently displacing the scar tissue adhesions through 20 hours of massage done over the course of 5 days.

Following a full week of this, I was able to get off half of the Oxycontin prescription my transplant doctors put me on to deal with the discomfort and pain. I then relocated to L.A. where I was able to begin my physical rehabilitation with a personal trainer to help build up the muscle that I had lost over the past year and a half. It was not until a year later in November of 2012 that another complication occurred. This time I was having issues with my breathing. It got to the point where I found myself constantly having to sit down and to catch my breath. Whenever I tried to take a deep breath, I felt like someone had just punched me in the stomach, knocking the breath out of me and leaving me winded. My transplant doctors ordered some blood tests to be done. As soon as they got the results, they told me my bilirubin was very high and I had to go to the ER immediately.

I initially went to a small, local hospital, in a small city, that didn't have the ability to do the blood tests that University of Pittsburgh Medical Center (UPMC) had originally wanted, that would show them my Tacrolimus (immunosuppressive medication) levels. The next day the head of my Transplant Team at UPMC told me I needed to fly up there immediately so they could figure out what was going on. At first, I was going to take the charter flight that the hospital uses, but it was not available at the time. We ended up finding a commercial flight leaving from an airport an hour and a half away in Fort Myers.

Upon arriving at the ER at UPMC Presbyterian, they rushed the order for my blood work and scheduled an MRI before admitting me to a room in Montefiore. I was alone there, as my grandmother and her other two part-time caregivers were in Florida and my parents were in Mexico. The doctors said my platelets were in the single digits and that they were REALLY worried about me flying (especially commercial). Platelets that low are VERY dangerous, and they had to give me infusions of new ones, just to draw my blood for testing.

I ended up having to stay in the hospital with several IVs in my arms for 2 1⁄2 months. Though they could not find out everything that was going on, they did find that I had the flu, a fever, walking pneumonia, and 50% fluid in my right lung. However, when they drained the fluid from my lungs there was evidence of a yeast infection. This was either because they gave me a yeast infection or found the infection within the fluid.

Unfortunately, my parents were not able to fly up to be with me until after the first of the year in 2013. I was battling a lot of nausea and often could not keep much food down. When my parents finally came, I remember they had to be dressed in special plastic hospital gowns and dawn masks, as the doctors were afraid that the germs might kill me.

I remember everything was very taxing and quite scary for me. My father had to fly down to help my grandmother while my mother and I flew down to Sarasota, Florida on February 19. I started physical

rehabilitation, rebuilding my muscles and stamina once again.

However, in 2017 I was rear-ended while driving back from a movie to my place. I ended up having to see my doctor and a couple of specialists. It turned out I had 5 herniated discs in my neck, so I had to do special exercises at a rehabilitation center. I was dealing with a lot of pain and having trouble keeping a straight posture. I was having a lot more migraine headaches which made my daily life difficult. It felt like I was trying to just get through the day with the fewest amounts of issues, but not really enjoying life at the moment. After a lot of rehabilitation, swimming, and writing to get through. I decided to start taking Ballroom Dance classes again, which I continued during the half of the year I spent in Florida.

That being said, a few months later the pandemic happened and thrust me into isolation. I ended up going back down to Florida because my parents managed to get a vaccine appointment down there. Afterward, my dad and I constantly checked different websites to get an appointment to get the vaccination. The U.S. Government had put transplant recipients in the "high risk" category for COVID, giving them eligibility to receive the shot. Sadly, Florida regulations had not caught up to the Federal Government at the time. This made getting an appointment for a shot much more difficult. When I managed to get an appointment, it was canceled because according to Florida's system I was not eligible yet. Finally, one pharmacy told me if they had anything left at the end of the day, they would call me to come down, since they had to cancel my original appointment. When I got the call, I received my first shot of the Moderna vaccine. I had the basic side effects such as arm soreness, little stiffness, and extreme fatigue, for a few days. The second shot did not give me any issues other than soreness and a little fatigue.

That August, I got my 3rd vaccination shot in North Adams, Massachusetts and had symptoms similar to when I had my first dose. I was the first person to receive their third shot at my Walmart Pharmacy. By the end of December, I got my 4th "Booster" shot down in Orlando (though this was a half a dose, and not a full one like the first three I had, per the rules for immunocompromised people). This time I didn't get any symptoms.

Being in isolation and worried about the vaccine situation gave me restless nights. I did not have many in-person interactions with people outside of my parents, no matter their vaccine status. I could not go to the studio and ended up trying to use Zoom in order to take my private dance lessons as well as group dance classes virtually. Whenever I was having a bad day or was in pain I would often share what I was dealing with in one of the MSUD Zoom meetings. I was super appreciative of the good wishes for a quick recovery from everyone. These Zoom meetings were started by Susan Needleman at the beginning of the pandemic to be able to share what we're dealing with and get suggestions to help with all sorts of issues. We even discussed our past experiences to help the younger patients benefit from them. These two interactions became the savior of my sanity and kept me from spinning into very deep depressions.

For a couple of months, I was able to go in person to my dance studio. It was great seeing everyone and being able to have an actual "person" as a dance partner again. So when I had to go back into isolation following the new year in 2022 (when there was a spike in COVID-19), I was sad. I was back to being a virtual dance student. My family also had some virtual reunions using Zoom during the pandemic. This was such a fun experience because I got to see people in my family who live in the Midwest, the Pacific Coast, and even in different countries.

My dance lessons always gave me something to look forward to and enjoy. I kept creating new ideas for dance choreography for my own performances or projects I wanted to do in the future with everyone. Seeing how much fun they were having made me jealous. I couldn't wait to return to the dance studio in person again and try dancing with my fellow students and my many instructors. And they couldn't wait to have me back in the studio.

The promise of being able to return to my dance studio at some point in the near future keeps my hope right on the horizon! ■

NEVER GIVE UP

By: Joel Felsenstein, Age 30, Classic MSUD

Hi everyone. Since my last entry Achieving the Impossible in the Spring 2020 newsletter, I have been doing a lot with my business ventures and expanding to the U.S. This year has already been an extremely busy time with meetings happening almost daily and getting everything moving. The first business will be concentrating heavily on general masterclasses, online courses, one-to-one private coaching, podcasts, and music to name a few, and I am incredibly excited to share with you all in this article. Aside from working on two business ventures and having countless meetings, I am still also actively promoting autism awareness and how everyone with it is totally unique and no two people are the same, it's the fact that we are all unique that makes us who we are.

Since the pandemic began, I have been involved in some new music projects and also been signed and endorsed by a few drum manufacturers. I am releasing my first single too this year and also launching my first business product and service within the music industry by going to view and potentially purchase a residential recording studio. On March 27th, I am going down to the southwest of the U.K. to view a residential recording studio that has a tourist side attached to it as well. This will be for the business I will be opening up in the U.S., too, that will work on the removal of the stigma and stereotyping behavior surrounding autism in the workplace, corporate world, and society. The setting of the studio we are viewing encompasses everything that my business stands for which is also our tagline: where creativity & uniqueness meets imagination.

I started driving lessons in 2020 and had my first lesson but then the COVID lockdown came. Now in 2022 my lessons have been going well and I am preparing for the U.K. driving theory test.

Before the pandemic stopped everything, I was planning my U.S. trip and then I had to suspend my plans, but the plans now have resumed for 2022 travel. My official website that will have online music

courses, recorded music, events, podcasts and more will be also launching later this year. It has been in the works since 2020 but due to some technical hold ups and legal issues it had to be pushed back, but now we are on track for launch.

I now teach music lessons both online and in person. I am passionate about helping others achieve their goals and aspirations, and also help them turn their dreams into reality. I am also a videographer and editor for a YouTube Channel that my new friend runs. Also, I have my own podcast shows, one for my business called RockstarX and for another other business of mine, Felsenstein Enterprises.

I look forward to helping everyone to achieve their goals and make their dreams a reality.

I was taught this motto by my mentor David Meltzer: Make a Lot of Money, Help a Lot of People & Have a Lot of Fun. This motto has always worked for me ever since I adopted it. Now, I want to pass it on to all of you.

Never give up on your dreams, aspirations, or goals. Always Work Hard, Play Hard, Dream Big and you will succeed in anything you put your mind to. Because you too can also Make a Lot of Money, Help a Lot of People & Have a Lot of Fun ■

RIVER'S STORY

By: Robert Cole II, Uncle, River 8 months old, Classic MSUD

Just before midnight on August 27th, 2021, my nephew River Cole was born in Oklahoma City. The entire family immediately fell in love with him. My brother Ridge Cole and his wife Sadie were excited to introduce River to his new home, and within a few days, River was released from the hospital. At first, everything seemed to be normal. River's first doctor's appointment didn't reveal any troubling complications, and aside from some jaundice to his face and chest River appeared to be a completely healthy baby boy.

Five days after his birth, Sadie received a call from a metabolic newborn screening nurse at Oklahoma Children's Hospital. They had found something in River's screening that appeared to be "a little off" and the results were indicative of a disease that can cause seizures or other health problems. The prospect of River having seizures was terrifying to the family and we were all very eager to learn more about this disease. After being advised to return to the hospital for further testing, health professionals proceeded to explain that River may have Maple Syrup Urine Disease (MSUD), a rare genetic condition that interferes with the body's ability to break down the amino acids in food. The staff provided an informational packet all about the disease before taking a sample of River's blood and urine. We were told that false positives do occur, so we were hopeful there was some mistake. Unfortunately, the blood and urine samples confirmed an imbalance in River's leucine level, meaning he likely had the disease.

Sadie received instructions to stop breastfeeding while the hospital awaited the results of more tests. During this time, River's dad Ridge started to notice the maple syrup smell indicative of MSUD but thought maybe the odor was caused by the formula they were using. River was sleeping quite often, but this didn't strike the family as something unusual or troubling. After another day, Sadie received a call from a nurse saying that River's bilirubin levels are normal and that light therapy would not be necessary to treat his jaundice. This was good news, but less than an hour later, the family received another call that shocked everyone. The nurse said River's leucine levels are up to 2,000, a potentially dangerous situation that could be fatal without intervention. This was the phone call that potentially saved River's life. Following the head geneticist's instructions, Sadie and Ridge rushed to the ER where River would need to stay under observation in the PICU for 3 to 10 days.

In the Emergency Room, Ridge and Sadie were immediately disoriented by all the confusion and commotion surrounding River. Sadie reflects on the night, "Everyone was hooking him up to machines and asking us if we realize how rare the disease is and how serious it is. They asked us if anyone in our families has MSUD, to which we kept replying, 'No, we have never heard of it.'" Several nurses even told Ridge and Sadie they were also unfamiliar with

River at 1 day old

MSUD and needed to research the disease further to give River the treatment he needed.

Before long, the staff moved River from the ER into the PICU where he remained for 6 long days. During this time, the doctors and nurses developed a feeding regimen for River, and this seemed to have a very positive effect. Sadie recalled, "I started to see a personality come out, he wanted to eat all the time and he ate very fast. We finally got to know each other over those 6 days and though I was still overwhelmed and scared, I was even more in love, and fall more in love every day."

It took a while for the family to get accustomed to daily testing of River's urine for ketones, not to mention keeping up with River's regimented dietary restrictions. Now, navigating River's medications and formula preparation has become second nature for the family. River is now 7 months old and weighs over 20 pounds. He continues to grow with each passing week. River's story illustrates that MSUD doesn't have to be a terrifying, insurmountable obstacle. River's grandmother, Jenny Godwin, finds that staying informed on the disease and connecting with other families coping with MSUD continues to be a source of strength: "Read everything you can find. It's a scary diagnosis and it sounds like the end of the world at first, but there's a great community out there with advice and support and you'll soon realize it's very manageable."

Ridge and Sadie found additional guidance through a Facebook group, and after talking with other parents of children with MSUD, their confidence in managing the disease rapidly grew. The entire family came together to show their support, and before long, coping with Maple Syrup Urine Disease was no longer such a scary, uncertain situation. However, River's success story is due in large part to the many doctors, nurses, and other healthcare professionals who came together to help. These people include River's metabolic doctor, Dr. Danielle Demarzo, and River's dietitian, Ashley Ethiedge. Other individuals our family would like to thank include Dr. Anne Chun-Hu Tsai, Michelle Peters, RN, and the entire staff at OU Children's Hospital here in Oklahoma.

Ridge, Sadie, and River today

River's other grandmother, Sami Cole, feels a tremendous amount of gratitude for the dedicated support provided by so many, both inside and outside the hospital: "It really does take a village to manage MSUD and we're just so thankful for all the help we've received. Everyone really came together to provide us with the education and resources we needed to navigate MSUD. River is simply thriving now and we couldn't be happier." ■

EVENTS

WHAT IS IT LIKE TO BE "OLDER" WITH MSUD?

By Susan Needleman (Age 31, Classic MSUD) and Amber Raye (Age 35, Classic MSUD)

On Saturday, February 5, 2022, a group gathered on a virtual Zoom meeting with 5 panelists that were either late 1st or 2nd generation of MSUD patients whose ages ranged from 43 to 53 years of age. The topic of discussion was what it is like to be "older" with MSUD. Even though we are "younger" with MSUD, listening to the MSUD panelists definitely brought up some memories of growing up for us. It should be noted that the information in this article is based on personal experience, not research. All disclosures said during the meeting were from personal experiences of those with MSUD speaking.

Back when the panelists were growing up much less was known about MSUD and the treatments that they as patients needed in order to thrive. What these individuals went through was drastically different from what most younger MSUD patients are experiencing today, as these "older" people have paved the way for newer generations. It is important to remember when something happens to one person or generation it does not mean that it is likely to happen to another person in the same way. Everyone has their own journey.

The first topic of discussion was regarding the various changes in formula throughout the panelists lives, which included the types they have been prescribed, method of receiving, preparation, and even storage. A majority remembered having to consume some form of formula from birth to teen years. Some even remember their doctor, theirselves, or their caregiver, having to add each ingredient separately, as it was not commercially made or sold. Once they transitioned into their adolescent years most of them had to switch formulas. They reminisced about the challenges surrounding storing the formula properly such as during travel and at school, as well as getting the smell out if it spilled on yourself or a surface. It is important to remember that back when they were younger there were no digital scales like there are nowadays. Some used balance scales. You could not eliminate the weight of the food from the dish, so they were forced to do the math to determine how their food or formula powder weighed. It was not until later on that the invention of the scoop spoon became more common in assisting with measurements.

The next topic that came up was the discussion of low protein foods, which seemed very relatable across the board. It seemed that everyone was in agreement that back when they were younger the quality of low protein food was poor. Not to mention the lack of options available versus today and ability to locate options at a local grocery store. Some even remember only being allowed to eat fruits and a few vegetables, but as much as they wanted of them. Exchanges did not exist yet and leucine amounts of foods were unknown. Today, it is no longer the traditional singular options of potatoes, rice, orange juice, saltines, and Fruit Roll-Ups. Now you have options and more flexibility in how to cook them. We all discussed the foods that were delicious back in the day or the ones that were absolutely disgusting that have since been discontinued such as the MSUD version of Jell-O, Herb Sauce, and bread in a can.

However, when it came to their blood leucine levels it seemed that the panelists all had different experiences. Some used to keep them higher because the doctors did not know that keeping them low was safer, while others kept their levels lower because their doctors did not know if they could safely have them higher back then.

Some of the other experiences that the panelists reported having are:

- There was a consensus among the panelists that stress does affect their levels and with the stress that comes with being an adult, it can be challenging to maintain lower levels.
- There was a consensus that most of them have experienced depression or anxiety and sometimes both.
- Sometimes as adults they struggled with knowing when their levels were up and their mother was often the one to know before them.
- They also observed that it takes them longer to heal an injury, like a broken bone.
- Some reported that they felt like they have weaker bones than those without MSUD.
- No one reported having other serious medical conditions as they got older.
- Many reported having experienced arthritis early, with half experiencing it before college
- Some reported muscle cramps when levels were low.
- Everyone agreed that they started experiencing aches and pains earlier than a non-MSUD person.
- All panelists find that their hands stiff up at times.
- No one reported having to change their calorie intake as they aged.
- A common problem that was reported among the panelists is dental problems from all the formula and sugar they have.
- Some reported low energy throughout life, while some started in their 20s or later.
- Some have early signs of eye diseases that non-MSUDers typically do not see until later in life, but each had signs of different ones.
- The one panelist who had received a transplant, 15 years ago, reported that at the very least, for a few years following their transplant, that they still had the maple syrup smell at times.

Tips and advice for younger generations from the older generations panelists

- Advocate for yourself especially when you are in the hospital.
- Do your own research about your health.
- Carry around an emergency letter from your clinic.
- Talk to others with MSUD. ■

MSUD VIRTUAL MEET-UPS NOW DIVIDE TEEN/ADULTS AND CAREGIVERS INTO SEPARATE MEETINGS

By Susan Needleman

There are a lot of people in this world, but few who have MSUD or 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 a couple of years ago, I started MSUD Virtual Meet-Ups. During these monthly meetings we talk about the struggles and successes we have had caring for MSUD, hear each other's stories, share what is going on in each other's lives, or just sit, listen, and take it all in.

In recent months I started to separate the attendees into two separate meetings, one for teens and adults with MSUD, and one for those

who care for someone with MSUD. With each position coming from different angles it started to make sense to divide them, which also has allowed for more time in the meetings to discuss additional topics.

The teen/adult meetings are usually held the first Saturday night of each month and the caregiver ones are held every second Saturday night of each month. If you would like to attend please email me at susanneedleman.msud@gmail.com. ■

HELP US TEACH PHYSICIANS ABOUT RARE METABOLIC DISEASE

FACT! Teaching about metabolic diseases (like the different types of Maple Syrup Urine Disease) in medical school and residency programs is poor.

FACT! Most patients live and die without a diagnosis being made, especially when the disease presents in adulthood.

FACT! Patients cannot access effective therapies **unless a proper diagnosis is made.**

FACT! The sooner a diagnosis is made and treatment begun, the better the outcome.

WE NEED YOUR HELP!

We at VMP Genetics believe in the power of "patient-teaching" and are bringing patients and families into lectures and presentations – at conferences and in the classroom. While doctors teach facts, patients tell stories. Story-telling is a more compelling teaching method with better recall over time than didactic lecturing. We also believe that doctors are more likely to make a diagnosis if they have already seen a patient and heard her/his story. Story-telling can be live or taped…

WE ARE LOOKING FOR…

- Patients and/or family members who are interested in telling their stories in local medical classroom settings… We are developing a Patient Teacher Registry. If a medical school faculty member is looking to introduce the patient story in a teaching session, the Registry can tell him/her if there are patient-speakers in the area and what diagnoses they have.

- Patients and/or family members who are interested in having their stories videotaped… As we secure funding, we are interested in recording stories that reflect the broader patient experience. The more variety in the stories, the richer the learning potential.

- Videos of patients and families telling their stories… A 5-or 10-minute clip can be downloaded into a lecture about that disease or relevant biochemistry to enhance the learning potential of the session.

Please help us in our efforts to raise awareness of metabolic disease through this innovative educational outreach to the medical community. For more information about this project… please contact us at PatientTeacherRegistry@vmpgenetics.com

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

The Board of Directors of the MSUD Family Support Group thank Susan Needleman and Amber Raye for their work as co-editors of this newsletter.

WANT TO SHARE YOUR STORY?

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