WWW.MSUD-SUPPORT.ORG

MSUD NEWSLETTER

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MSUD REGISTRY: WHAT WE HAVE LEARNED FROM YOU

Have you participated in the MSUD Registry with Sanford CoRDS yet? If you are one of the 145 people who did, thank you! A few people registered but didn't complete the questionnaire. Here's what we have learned so far from 117 responses:

- 87% of respondents have Classic MSUD
- 55% have been hospitalized 5 times or more
- 13% are seen by their metabolic team monthly or quarterly; 73% are seen semi-annually or annually
- 53% receive the results of routine blood tests within 48 hours
- 24% report having received a diagnosis of an anxiety disorder
- 41% report strict compliance with diet
- 23% report out of pocket expenses related to MSUD of more than $2500 annually
- 14% report receiving individual assistance at school
- 32% of adults report living with their parents

Please help us learn more by registering if you haven't already done so. We need to hear from as many people as possible, including those who are transplanted and those living in other countries.

Do you prefer to respond in Spanish? We have you covered! Contact CoRDS by calling 877-658-9192 or emailing them at cords@sanfordhealth.org to request Spanish language questionnaires. ■

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

By Butch Foster and Ed Fischler

The Mays Family

The MSUD Family Support Group again participated in the Penn Medicine / Orphan Disease Center's Million Dollar Bike Ride (MDBR). The Penn Medicine Orphan Disease Center (ODC) was established to promote the development of therapies across a broad range of orphan diseases, defined as those affecting fewer than 200,000 individuals nationwide. The ODC sponsors and promotes the annual MDBR to help organizations raise funds for pilot research grants for the rare disease represented by each participating team. Due to the pandemic, this year's event was again "virtual," with riders completing their rides in and around their neighborhoods on June 12th.

This year, fourteen families responded to the need for riders and campaigners for MSUD. These included the families

Million Dollar Bike Ride continued on page 3

Inside This Issue:

Sections

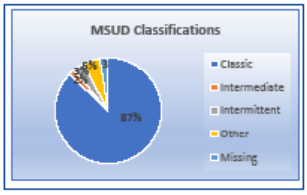
Key Findings From the MSUD Registry

MSUD Family Support Group

Results based on 115 responses

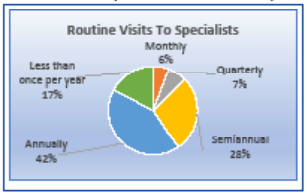
MSUD Classifications

A clear majority of respondents (87%) indicate that they have "Classic" MSUD.



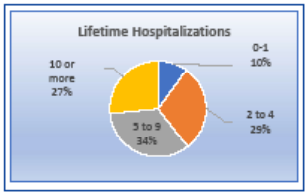
Routine Visits To A Metabolic Specialist

Almost three-quarters of MSUD patients visit their metabolic specialists twice or once a year.



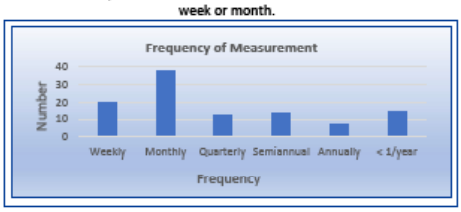
Lifetime Hospitalizations

Most MSUD patients have been hospitalized at least five times in their lifetime.



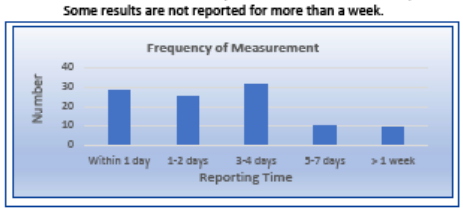
Frequency of Blood Amino Acid Measurement

Most MSUD patients measure their blood amino acid levels each week or month.



Timeliness of Blood Amino Acid Measurement Results

Most measurement results are reported within three or four days. Some results are not reported for more than a week.



FROM THE PRESIDENT'S DESK

By Sandy Bulcher

The MSUD Family Support Group Board of Directors met at the beautiful Massachusetts home of board member Herb Foster and his wife Diane on July 16th and 17th. We are grateful for their warm welcome and generosity.

After our last in-person board meeting in the summer of 2019, none of us could have predicted the onset of Covid and how it would impact our lives. But despite Covid, the MSUD Family Support Group Board has continued working toward our goals of connecting and engaging families, progressing research to improve the lives of those with MSUD, and advocating to meet the needs of our community.

During the two day meeting, we discussed ongoing projects including the next MSUD Symposium which will be held in Lancaster, PA at the Lancaster Marriott on August 4-6, 2022. It will be hosted by the Clinic for Special Children. Mark your calendars now so you don't miss this event!

President's Desk' continued on page 3

From the Editor

By Karen Dolins

I have been editing this newsletter since 2004. At that time my daughter was 10 years old and we had traveled together to the MSUD Symposium in Atlanta, Georgia. I met wonderful people there, including Joyce Brubacher who founded this organization and had been serving as editor of the newsletter. She was looking for someone to take over as she was also involved in many other aspects of running our organization. I agreed.

Now, 17 years later, it is time for me to step down from this role. In addition to editing the newsletter, I serve as a board member and Secretary for the MSUD Family Support Group and am also our Research Lead. In this capacity I communicate with members of our Scientific Advisory Board, follow current research related to MSUD, and suggest future avenues of research. As you can imagine, this keeps me quite busy. I realize that it's time to focus on this critical area and let others take over the newsletter.

I'm very excited to announce that 2 adults with MSUD, Susan Needleman and Amber Raye, have volunteered to take over as co-editors. They have introduced themselves in this newsletter and have been helping with this issue.

You will continue to hear from me as I will provide the content related to research for this newsletter. I will keep you apprised of our research efforts as we strive to improve the lives of those with MSUD and search for a cure. ■

President's Desk continued from page 2

Also, check out the new and improved MSUD website (www.msud-support.org). See article in this newsletter for more details.

We partnered with Sandy van Calcar RD, PhD and graduate student, Elaine Sim, to produce a maternal pregnancy MSUD booklet. I am excited to let you know that it has been completed and is available on our website. If you would like a hard copy of the booklet, please reach out to me by email sandybulcher@gmail.com or phone 740-972-5619. We are grateful for all of their hard work on this project.

Our efforts to promote the CoRDS MSUD registry continue. Watch for more information including incentives for completing a survey. We are consistently told by researchers that a comprehensive registry is the single most important thing that we can do to advance MSUD research. We are working hard to accomplish this goal. Please participate in the registry and encourage others to do so, including those that have had a transplant.

To remain fiscally responsible, we scrutinized our finances and budget during our board meeting. We were able to identify four projects that we feel are the most important to fund.

These include:
1) registry/natural history studies
2) home leucine monitor
3) gene therapy
4) neurocognitive impact of MSUD.
These priorities will help guide our resources and activities in the future.

We concluded the meeting with a conversation about the importance of planning for our succession as many of the board members are "getting up there in years". If you are interested in becoming more involved in the MSUD Family Support Group, please reach out to me. For us to remain effective, we need both financial and personnel resources. Please consider fundraising, donating and /or volunteering.

A donation slip is included in this mailing. Your financial support makes it possible for us to continue our work. If your contact information has changed, please include that information in the slip and return it so our database is as accurate as possible.

As always, feel free to reach out to me any time at sandybulcher@gmail.com or 740-972-5619. ■

Million Dollar Bike Ride continued from page 1

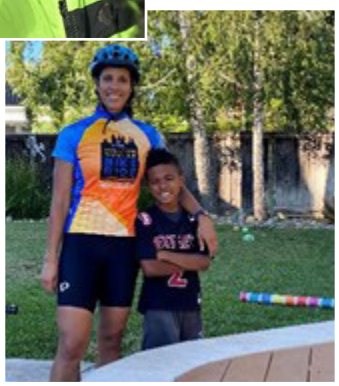
of Andrea Brien; Jordann Coleman; Susan Eliason; Edward Fischler; Herbert Foster; William Goodwin; Stephen Healy; Leslie Hirshfeld; Ashley Kelly; Doug and Taryn Kessel; Karen and Jerry Dolins, Michelle Flanagan; Susan and Adrian Mays; Susan Needleman; and Cristy Steinberg.
Let's again thank the whole team who helped to raise funds for MSUD research. Research to find better treatments and a cure for diseases like MSUD is a journey. We all want to improve the lives of families affected by MSUD. These families have done some of the heavy lifting necessary to fund the research to achieve these goals. This year, donations for over $59,000 was contributed to the MDBR campaign towards MSUD research. The University of Pennsylvania/ Orphan Disease Center has matched our amount with an additional $30,000, making the total amount raised over $89,000!

This was the fourth year of our participation in the MDBR. Since our participation began in 2018 we have been able to fund four research projects focused on MSUD. This year we will add to that total. The MSUD Family Support Group Board of Directors has directed the organizers at UPenn to seek proposals to fund two MSUD research projects:

- One project seeking better treatments for MSUD and
- One project directed towards the goal of finding a cure for MSUD.

The ODC issued a request for project proposals in August. Reviews of the responses should be completed by November and the selection of the winning proposal(s) will occur in December.

Again, thank you for your outstanding support of the MSUD community! ■



SUSAN NEEDLEMAN CLASSIC MSUD, AGE 31 NEWSLETTER CO-EDITOR

My name is Susan Needleman. I am 31 years old with classic MSUD and am the new Co-editor of this newsletter along with Amber Raye. I was diagnosed through newborn screening when I was a week old.

The test results changed many things for my family, some of which have continued to change, as my needs change, and I get older. However, the one thing that never went away was our subscription to the MSUD Newsletter. Even as a young child, I remember how excited I was when the MSUD Newsletter would arrive and how I wanted to read it from cover to cover. When I could not fully read the Newsletter on my own, much less understand it all, I liked knowing that all the contents within it were connected to me somehow. As I got older and could read and comprehend the contents of the Newsletter, I would read every word from personal stories, research articles, recipes, you name it. One memory that stands out was when I was ten years

Susan Needleman continued on page 5

AMBER RAYE CLASSIC MSUD, AGE 35 NEWSLETTER CO-EDITOR

My name is Amber Raye, and I recently accepted the opportunity to take on the role of co-editor of the MSUD Newsletter alongside Susan Needleman. I am 35 years old and was diagnosed with Classic MSUD at seven days old. The primary reason I took this position was to learn from each of my fellow MSUD peers by editing their stories and experiences. Throughout school, I have always been the peer or friend who would edit papers for those who needed them. It was the sophomore year of my BA at Ashford University where I discovered my love for editing. However, it was not until the last few years after interacting through social media with other MSUD families that I began to take on a more significant role within the MSUD community and provide readers with various resources that affect our daily lives.

Currently, I reside in a tiny town in Massachusetts with my father, fiancée named Chris, and three cats (Loki, Valkyrie, and Iris- Lullaby). I most recently began to work with a feline sanctuary where I help socialize cats that were abandoned, abused, semi-feral, special needs, or saved from death row. I also help manage a tiny bookstore, which I love since I am a huge bookworm.

I love cooking new low protein dishes and being creative. My all time favorite is low protein potato soup. It was a recipe that my great grandmother taught me while I was growing up that made me fall in love with cooking. Zucchini Pizzas is another creative idea for a low protein recipe dish that I learned a bit ago, made with zucchini, low protein shredded cheese, garlic, and a pinch of crushed red peppers. I am a strong advocate of finding new and innovative ways to spice up dishes to avoid eating the same things repeatedly. For example, I feel like most of us always turn to potatoes for an option when there is nothing else. So,for dishes like sweet potato cheesy hashbrowns, you can take one sweet potato peeled and some olive oil in a frying pan to begin to make home fries but add some shredded low protein cheese and garlic to the mix). Cambrooke tortilla wraps make a flatbread sandwich by cutting the tortilla in half using one or two Cambrooke cheese slices, then add some tomato, onion, or mushrooms. Be creative as long as it is within the confines of your protein allowance. Then cook it like you would a grilled cheese.

I look forward to learning about each one of you and working beside everyone on the Newsletter. I am excited to begin my journey as co-editor and work beside outgoing newsletter-editor Karen Dolins and Susan Needleman to continue bringing the newest information to your home. ■

Susan Needleman continued from page 4

old. I went to the Boston Symposium in 2000, and I had picked up some old newsletters that came out before I was born. I remember enjoying reading them and learning about what happened back then. Those became some of my prized possessions, and to this day, I still have them and every single one that my family has received since I was a baby. At times I even look back and reference them.

Fast forward to my adult years. I was always involved to one extent or another in the MSUD community. Once I graduated from college, I became more involved through connecting with families, participating on social media platforms, hosting virtual MSUD Happy Hours, and joining the board of the New England Connection for PKU and Allied Disorders, a regional support group. Since MSUD is an allied disorder of PKU, I quickly became the voice of MSUD in the group. I went on to become the Allied Disorder & Outreach Coordinator, which allowed me to work with MSUD families. I am now the Vice President of the group and am excited about collaborating with families who are affected by MSUD and similar disorders.

As time went on, I wanted to have a more prominent role within the MSUD community. When I saw the posting for the position of Newsletter Editor, I immediately jumped for the opportunity. English and editing is my strongest suit and I have always enjoyed creative writing. This allows me the opportunity to combine all my passions together! I am very excited to take on this new position and provide the MSUD community with resources through this Newsletter. ■

NEW MSUD LOGO

You may have noticed that the MSUD Family Support Group has a new logo. While updating the look of our website, we also thought it was time for a refreshed logo. The new logo provides a more modern look, but our emphasis on family and community remains consistent from the previous logo. Thank you to Michael Fischler for helping us revamp our logo. ■

MSUD Family Support Group

SUPPORT THE MSUD FAMILY SUPPORT GROUP

The MSUD Family Support Group is a non-profit 501 (c) 3 tax exempt organization and is run entirely by volunteers. We are dependent on donations from the MSUD community and others to support our work.

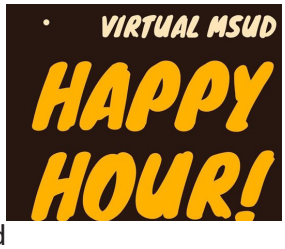
Your donation will allow us to publish a newsletter, put on a symposium, pursue research, and advocate for the needs of those with MSUD.

Donations can be directed towards general operations or research:
• The general operating fund supports symposiums, mailing and printing of newsletters, other communications, and professional fees.
• The research fund supports research projects deemed worthy by our Scientific Advisory Board and with approval of the Board of Directors. A list of past, current, and future research projects can be found on our website, msud-support.org.

All donations are tax exempt. Our tax-exempt ID # is 54-1556981.
Donations can be made by mailing a check to:
MSUD Family Support Group c/o
Dave Bulcher, Treasurer
4656 Winding Oak Drive
Delaware, Ohio 43015
or by going to the website and clicking on the donate button at the bottom of the main page.
We sincerely appreciate any donation you may choose to make. ■

MONTHLY MSUD HAPPY HOURS

by Susan Needleman



The pandemic brought a lot of new experiences into our lives. Some of them will go away as we get back to normal, but one that will not is the MSUD Happy Hours! The MSUD Happy Hour is an online Zoom get-together of MSUD adults and parents of children with MSUD, both transplanted and not, from around the world. It started during the early days of the pandemic when many of us were isolated at home, allowing us to connect, and not be alone.

However, it has evolved into something much more than just that. It has become a safe place for the MSUD community to come together and establish friendships, catch up after not seeing each other since the last meeting, discuss topics that we encounter within our daily lives, and have fun! It is a time for friends to come together, have formula and/or a meal with the group, or just talk. It allows members to obtain support, receive advice, share experiences, connect with others that have MSUD and not feel so isolated with it. MSUD Happy Hours are constantly growing with new members! If you would like to join the monthly Saturday night meet-ups, email susanneedleman.msud@gmail.com. ■

ADVOCACY UPDATE

By Jordann Coleman

The Medical Nutrition Equity Act was introduced in the House (H.R. 3783) and Senate (S.2013) this past 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 11 co-sponsors in the Senate and 42 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 does make 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).

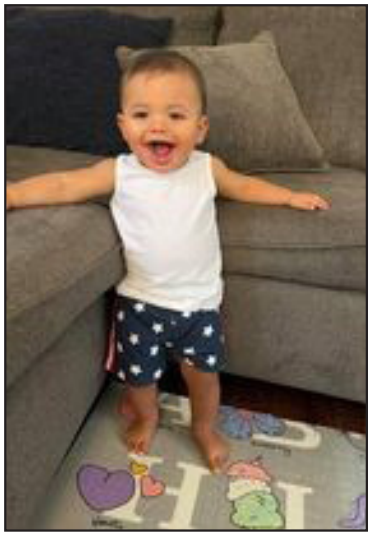
Please share with your friends, family & social networks and ask them to contact their Congressional members. Together we can make coverage for medical nutrition a reality.

In addition, The Newborn Screening Saves Lives Reauthorization Act passed this 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. ■

FAMILY NEWS

JOHN JESSE "JJ" MORALES CLASSIC MSUD AGE 1

By Shyanne Kelly, Mom



Within a week, I experienced the happiest day of my life, and also the scariest day. On May 29th, 2020, I gave birth to our perfect, amazing son, John Jesse Morales. We call him JJ. He had the biggest, most beautiful eyes, a full head of hair, and the cutest little grumpy face; he was just perfect. The day he was born was even more special because JJ is our rainbow baby. Not even a year prior to his birth, I was pregnant with another baby, but unfortunately I miscarried at only 11 weeks. JJ was conceived only 6 weeks after the miscarriage, so the trauma from losing our first baby was still pretty fresh. Throughout my pregnancy with JJ, terrified that we would lose another baby, I was just praying at least three times a day that God would protect my baby and keep him safe. When he was finally born, all that anxiety and fear washed away, and was replaced with so much love, joy, and peace. He came into this world a fighter, screaming before he was even pulled all the way out of my womb by Cesarean Section. The moment the doctor showed him to us for the first time, my husband John and I felt incredibly blessed and that the hard part was over, and our miracle baby was finally here. Little did we know our world was about to be flipped completely upside down.

We spent the next couple days in the mother baby unit recovering. JJ was having a hard time drinking. We tried breastfeeding but he would fall asleep almost immediately every time. We thought maybe he was just too comfortable on mama's chest, so we introduced formula and he responded well to that. Once that was taken care of and I healed up a little more from my Cesarean section, we were finally discharged. Since JJ had a hard time nursing at first, his weight was dropping a little more than his pediatrician would like, so we were told to make an appointment with his pediatrician for the following day, to check his weight again.

I told you about the happiest day of my life, now it's time for the scariest day of my life. Once we got home, we noticed the formula wasn't working for JJ like it was before. He was falling asleep while drinking, just like he did every time we tried to nurse. We went to his pediatrician's office for his weigh-in, where we were totally blind-sided. His doctor informed us that JJ's newborn screening came back with elevated leucine levels indicating he may have a rare genetic disease called Maple Syrup Urine Disease, a disease so rare that he would be the first person in our state (Alaska) to ever be diagnosed with it. John and I had never heard of this disease, and were terrified. I was already feeling stressed as a first time mom, but now I'm a first time mom to a baby with a rare disease? John and I both felt completely in over our heads. We did additional lab work to confirm the diagnosis, and brought JJ home where we waited for results. His doctor advised that we continue with his normal routine including formula until then, and to bring him in if he became so lethargic he couldn't eat.

Over the next couple days, JJ grew increasingly lethargic. It would take two hours for him to eat not even two ounces. We decided to bring him back in.

At birth, JJ weighed 7lbs 3oz. At 8 days old, he weighed 6lbs 4oz. He had lost almost a full pound in just a week. The weight loss accompanied with lethargy warranted an immediate admission to the pediatric intensive care unit. None of the doctors in the PICU had ever treated a patient with MSUD, so JJ's case was overseen by a specialist in Portland, Oregon. His leucine level was rechecked, and we discovered that they had skyrocketed to over 1,600 (normal range is between 70 and 300). A day later when it was officially confirmed that he had MSUD, JJ was given a new formula specifically for babies with MSUD. Once he was given that formula, he finally opened his eyes for the first time in almost 48 hours.

As happy as we were in that moment to see his big, beautiful eyes again, we would soon realize that was just one small victory. The next three weeks were hell for my poor, sweet

newborn baby. He had Covid testing, a spinal tap, ultrasounds, X-rays, a nasogastric (NG) tube placed, I can't even tell you how many times he was poked for an IV because his veins were so tiny they kept blowing out, he had labs drawn daily, he met with countless doctors and specialists and therapists and intensivists, and so much more. He was just poked and prodded and chained to his bed with all the leads and wires. Throughout all of this, his dad and I never left him, we stayed right by his side; my 6'7" husband slept on the tiny couch in the room, and I slept in the chair. John only left to go to work, and would come back to the hospital right after. I would only go home to shower once every 3-4 days. I have never met anyone as strong as my son. He wasn't even a month old yet and had already gone through more than some people will go through in a lifetime.

Finally his levels started coming down. He was drinking more and more from a bottle instead of his NG tube, and he was so alert and happy again. After those three LONG weeks, JJ was finally discharged and we brought him home again, but would need to come to the hospital daily to get his labs drawn.

It was quite the learning curve: how to calculate leucine, how to mix his formula, how to spot behaviors indicative of rising levels, and honestly I texted JJ's dietitian more than I texted my husband those first couple months. For the first six months of life, JJ had terrible acid reflux from his MSUD formula being so thin and would scream all day and all night. The acid reflux got better once we introduced solids, but then the teething started getting out of control. In one week he cut SIX teeth! Every time he's in pain or stressed, his levels go up, so we've done countless sick-day diets. But with the help of my husband and our great support group (friends, family, and the amazing people I've met through the MSUD support page on Facebook) we got by.

Today I am very grateful. JJ is now 14 months old, and if I didn't tell you, you probably wouldn't know that he has MSUD. He has come so far. He used to be a tiny, frail, lethargic, 6lb 4oz, 8 day old baby who wasn't even on the weight chart, and now he is absolutely thriving. Today, he is a big and strong 26lbs 4oz little boy- putting him at the 95th percentile for his age (for both weight and height)! He is brave, smart, curious, sweet, kind, handsome, playful, and best of all, a total mama's boy. John and I are incredibly proud of our son, not just the progress he's made, but also the awesome little boy he's become. Most importantly, JJ is happy! He does not let this disease define his life, he still does all the things a "normal" toddler would, like scaring the hell out of me and climbing dangerous things he knows he's not supposed to, with a devious smile on his face of course. We take him out to dinner, go on adventures, bring him to new places, and we plan on bringing him on a vacation sometime soon! If you told me this would be the life we would live with him 14 months ago, I wouldn't have believed you. I was so scared, worried, and overwhelmed thinking about what his future might hold. I felt totally in over my head, alone, and frankly unqualified back then. But as a sweet woman I met through the MSUD support page told me: "God made us their mothers because He trusts us to take care of them." Every day I do the best I can for my son, and I hope that I can be the mother he deserves because he's quite a special boy, and I love him more than life itself. He deserves nothing short of the best.

If your baby was recently diagnosed, I know it all seems so scary and overwhelming now, but over time it will get easier to manage, I promise. Just take it one day at a time, count your blessings, no matter how small they might seem, and don't be afraid to reach out and ask for help, or even just vent. I don't think I could've made it the past 14 months without the incredible support system we have today. I will always be here for you, like others were there for me and my family! ■

MSUD PREGNANCIES: SUCCESS STORY AND ADVICE

By Rachael Pokrovski (Ennis)

Classic MSUD, Age 37

MSUD and pregnancy is always a challenge, and essentially new ground. When I was growing up, I did not know if having children was a possibility since I had classic MSUD. The documented cases that existed when I had my first child allowed us to create a plan with my team at Massachusetts General Hospital (MGH). Having a plan helped us feel more confident. We had plenty of time to plan, too, because it took us 2 years to get pregnant, and this was after some failed IUI (intrauterine insemination) efforts and a miscarriage.

I was 28 when I got pregnant with our oldest, Rosalyn. The normal maladies of pregnancy such as morning sickness all came and made pregnancy difficult. I am not sure the morning sickness affected my levels that much, because for me it never really kicked in until week nine or ten, and if I did throw up I would just drink another MSUD shake. . . .I will say week 10 was tough, because I needed IV fluids due to throwing up from morning sickness, which caused me to be both dehydrated and short on calories.

My normal leucine tolerance is 8-10 grams of protein (without any from eggs, dairy, meat, etc.), but with the pregnancy it went up to about 20-30 grams in the third trimester, and could be from whole protein foods like beans, meat, cheese, and milk. My leucine tolerance was also slightly higher with my oldest, because she was a much bigger baby born at 8 lbs 14 ounces, than with my younger who was born at 6 lbs 6 ounces. You cannot dictate how they grow really; some babies need less/take less and that's just the way it is. I think the third trimester has traditionally been the toughest for me, even though I could eat more different foods than normal, not necessarily more food, but I could tolerate a wider variety of foods---such as small quantities of milk, meat, higher protein grains, things like pasta and quinoa, and peas that I do not eat when I am not pregnant because of MSUD. I was physically very uncomfortable and pretty nervous. I also had a lot more heartburn and digestive issues with new foods I was trying.

During this first pregnancy I stubbornly insisted that I try labor, even though I would need IV fluids that would increase the risk of my baby being born with low blood sugar. At 39 weeks and after 34 hours of induced labor I ended up having a c-section at 10:27pm on 08/27/13. My beautiful baby girl Rosalyn was born with low blood sugar and in the NICU for a few days, but was otherwise healthy (I could have saved myself 34 hours, and not been separated from my baby, it was very hard to have that happen even though she would be ok). I was in the hospital on a floor removed from the maternity unit for two weeks to stabilize my levels (they peaked at about 900 µg/L), and since Rosalyn was discharged from the NICU I had to have supervision 24/7 to be with her during my recovery, which actually made it harder for us to bond. Looking back, I probably would have preferred they do peritoneal dialysis to lower my levels more immediately, and/or increase the calories more to give me the energy I needed to recover and transition to nursing. I was not able to develop a milk supply, unfortunately, and dealt with some postpartum depression as well.

With my second baby, I thought I was more prepared and knew what would need to happen. I normally test my leucine levels a few times a year, more frequently if I feel like they are off, but during the pregnancies I tested them once a week. I would drive into Boston Children's Hospital and get results the next day, often having them drawn on Saturday mornings and getting results on Monday. I knew I was pregnant at about 4 weeks with each pregnancy, because we were trying and checking every month that my period was late. Even though I knew my levels needed to be low during my first trimester, and I was trying to keep them low, I had issues with my levels during week 6. The first 6 weeks are critical, and if you get sick or are not feeling 100%, and levels elevate, it can cause problems. It's important to do whatever you can to:

a. know you are pregnant
b. keep levels low

My leucine levels were just under 700 when I got a blood pregnancy test with my OBGYN during the 6th week to confirm my second pregnancy. This is right around the time the heart develops, but also a time when many women do not even know if they are pregnant. My elevated levels may have caused her to have a common heart defect called Tetralogy of Fallot, which was picked up on ultrasound at week 22. There is no way to know for sure whether my elevated levels caused it (since there are so few MSUD pregnancies). The same defect occurs with PKU moms. Both leucine and phenylalanine can be teratogenic (cause developmental problems in a fetus), so I believe it may have been a factor. PKU moms usually need to have a blood phenylalanine level over 900 to have that kind of impact, but that threshold may be lower with leucine, based on my experience.

I also had gestational diabetes during this second pregnancy, which was mostly managed with insulin. I had to monitor my glucose levels with at home finger pricks throughout the last half of the pregnancy. I had to have an insulin drip along with standard MSUD IVs, while delivering Elaine. Elaine was born on 04/26/19 at 37.5 weeks, and despite her heart condition she has been thriving and developing normally, reaching all milestones at or ahead of schedule. I had a scheduled C-section, and even though Elaine needed to be in the NICU for a night or two (for low blood sugar, and some minor

apnea), we shared a room for two weeks at the hospital, and thanks to very prepared teams at Brigham & Women's Delivery Unit and Boston Children's Hospital's Metabolic Department my levels never reached much above 500. They provided considerably more calories to support nursing/milk supply for pumping, which really helped. Elaine had open heart surgery and a full repair at one month old. She had some Occupational Therapy through her first year, which is standard for heart warrior babies, but she graduated out of the program at 18 months, since she was right on track. She just turned 2 and is ready for daycare in the Fall.

We are lucky to have both of our children and feel blessed that they are overall healthy. Elaine's prognosis is great and she most likely will never need surgery again, just possibly a catheter someday to widen an artery if it doesn't widen on its own as she grows.

We feel very fortunate, and just want to reiterate that it's so important to keep levels low in the early stages of pregnancy. Leucine tolerance does increase at the very end of trimester 2 and more considerably in the 3rd trimester, after the baby's stomach develops, and the baby grows. Having enough calories, water, and a prenatal vitamin, as well as some regular, safe exercise is super important as well.

Having our second baby took us 3 years of trying; the cause of our infertility was unknown and I have no way of knowing if my levels need to be really well controlled to make it work, or if I need to weigh less to make it easier (I was 170 lbs when I got pregnant with Rosalyn and 200 when I got pregnant with Elaine). I also do not know if MSUD makes it more difficult in any way or it's just me/us, but each time we didn't give up despite the long road we knew was ahead.

I just wanted to highlight some of the challenges, as well as the positive outcomes of this process, and to show that not only is it possible, but it is immensely worth it!!! I wouldn't trade my kiddos for anything in the world!!! Thank-you to both my original team at Massachusetts General Hospital and my new teams at Brigham and Women's Hospital and Boston Children's Hospital for making this happen as safely as possible.

Editor's Note:
For more information about MSUD and pregnancy, please see the MSUD and Pregnancy booklet which is posted on our website. If you would like a hard copy, please contact Sandy Bulcher at sandybulcher@gmail.com. ■

THE MSUD FAMILY SUPPORT GROUP HAS A NEW WEBSITE!

Have you visited www.msud-support.org recently? If so, you may have noticed a new look. Our website has been updated to make it easier for you to find the information you are looking for, making it your "go-to" resource whether you are a new parent, adult with MSUD, professional, or researcher.

Headings include:

- What is MSUD?: Find out about diagnosis, treatment, liver transplant, and more.
- Research: Get updates on research projects aimed at improving the lives of those with MSUD.
- Take action: See what you can do to advocate for those with MSUD.
- Resources: Find a clinic, get nutrition information, and access the new MSUD Pregnancy Guide.
- News and events: This is where you'll find our newsletter, symposium information, rare disease conferences, and information on the Million Dollar Bike Ride for rare diseases.
- Who are we? See a list of our medical and scientific advisors, read personal stories, and find rare disease organizations we work with.
- Contacts: Want to help? Sign up to volunteer here!

The website also makes it easy to donate to our organization by having a button on each page. Donors can opt to fund general operations or contribute to our research fund.

Take a look today! I guarantee you'll learn something you didn't know about MSUD and our organization! ■

FOOD & NUTRITION

THE FORMULA DIARIES

By Amber Raye
Classic MSUD, Age 35

As individuals with MSUD, we tend to have love and hate relationships with drinking our formula, or what I like to call 'The Bog of Eternal Stench' (Labyrinth reference) or whatever term of endearment we have chosen to call it over the years. All of us have encountered at least one headache when placing our formula orders. It might be that the warehouse is out, the person who is taking the formula order is not familiar enough with our formula that they make an error in the order form, or the warehouse packers pack the completely wrong formula altogether. It is imperative as we deal with these errors that we continue to be vigilant in ensuring we receive the right formula for MSUD, as a simple mistake that is missed could potentially cause catastrophic issues to an individual with MSUD.

Some of you may have heard your doctors touch upon a case years ago where a classic MSUD patient had received the wrong formula which triggered a metabolic crisis that continued throughout the year. That patient was ….me.

It is critical as patients that we are aware when receiving formula or prescriptions for MSUD to ensure that we are getting the correct items in the order. Here are a few tips to ensure accuracy have helped minimize mistakes that I have learned over the years.

1. Make sure you talk to a rep: Have them read back what items you are supposed to be receiving, as this will ensure that the representative was actively listening while you were placing your monthly refill.
2. Always get their name at the beginning of the call: This will give you a reference point for a delivery issue or if the package is late.
3. Ask them when it is expected to arrive: get a tracking number if you do not already have one. It helps to eliminate potential errors in delivery to the wrong house and gives you a road map if you need to reference it later.
4. If issues keep occurring, ask for a manager and see if someone from the management team can handle your monthly reorders to ensure accuracy. Never be afraid to ask to speak to a manager if there are issues.
5. Try to save a few cans each month without reducing the amount of formula you are supposed to be consuming. Even if it is 1, it will help immensely in case of emergencies. If this is not a possibility due to limited supply, please get in touch with your metabolic team to help develop a plan if needed.

It is important to understand how these mistakes may impact us and how to identify them when we are receiving our orders. We are human, after all, and these mistakes, though not common, do happen occasionally.

But how do we spot these mistakes? And how can we minimize the likelihood of them occurring? First, be aware of what the label looks like and what the writing on the can or container says ((i.e., use, flavor, ages it is used by, etc.), colors, and images. This may seem silly to someone without MSUD; I mean, our doctors write the prescription, so why does this happen? Mistakes happen all the time. It could be due to the packer not paying attention to the order they are packing or being burned out and tired. Someone not aware of the differences in the formula we receive and others they may carry, or lack of understanding of rare metabolic diseases might not pay so close attention.

Unfortunately, these mistakes could ultimately leave an individual with MSUD in critical condition if not caught before consummation and fixed accordingly. For me, the nightmare occurred when I was seven years old, about 28 years ago. I was supposed to be receiving the MSUD Maximum at the time. However, the company used the wrong formula with subtle differences in the label, they gave me cans with blocks instead of circles and was not for my age. This resulted in a year-long battle for my life (i.e., brain edema, significantly high leucine levels, hallucinations, hospitalizations, endless blood drawings, and seizures). It was not until another MSUD specialist had heard my case from another state and had come to my hospital bed that we got to the bottom of my case. He ultimately saved my life by finding out it was the wrong formula and got it fixed immediately.

So what do we do when there is an error in our formula order? The first thing is to not consume it if the formula does not look right, (i.e., the label doesn't match what last formula cans or packets have in the past, notify the company who shipped it immediately. Next, inform the on-call or metabolic doctor at your clinic immediately to let them know, and if you have extra cans from previous orders, check them to make sure that they are correct before consuming. Lastly, if at risk of running out due to this error, let the on-call metabolic doctor or someone else in your metabolic team know ASAP so they can ensure that you have the resources to obtain your formula until the error is addressed. I cannot state this enough to be on top of the company if this occurs, even if this means asking to take it to the manager or higher-ups. Please never be afraid to ask questions if you are not sure.

My goal with this article is to educate MSUD families internationally on how to advocate for themselves in ordering their formula and handling issues or concerns that arise during the ordering and delivery process. ■

HAILEY'S SWITCH TO MSUD EASY TABS MSUD THIAMINE RESPONSIVE, AGE 20

By Michelle Petty, Mom

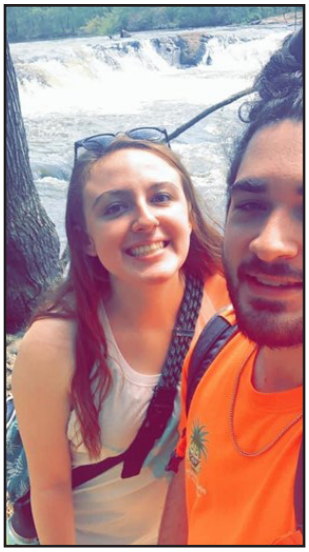
Hailey is a 20 year old MSUD patient that is Thiamine responsive. She had been taking Ketonex formula since her diagnosis at 2 weeks old. We have never had any issues with her consuming the prescribed amount of formula. She's always been willing to drink it with ease and even craved it from time to time. Some of the biggest drawbacks to the formula, in Hailey's experience, was traveling with a large quantity of cans, storing refrigerated components, and having to weigh and measure out her prescribed amount each day. She was an avid soccer player for most of her life and found that the high calorie content in the Ketonex formula served her well with the level of physical activity she endured within a typical week. As she has gotten older and her fitness demands have decreased now that she is no longer playing competitive sports, she has found the higher calorie consumption to be more of a concern than a benefit.

It wasn't until recent years that Hailey started asking for alternatives to the Ketonex. She attended metabolic camp at Emory University in Atlanta, GA for several summers. She was one of very few MSUD girls that attended, most were PKU patients. Hailey became very interested in the possibility of MSUD Easy Tabs which is virtually MSUD formula in pill form. At the time it was being produced she saw many of her PKU friends coming off of formula and taking the PKU Microtabs, which are very similar.

She reached out to her metabolic team a year ago to request that she be allowed to explore other formula options with a lower calorie intake. After trying several brands and finding it difficult to stomach most of them, they finally came around to the idea of the MSUD Easy Tabs. She first started with a trial of the medical food tablets to see how her levels would respond and if she could tolerate a high quantity of pills in a day. Hailey has to consume a large number of tablets each day, as 11 tablets only have 10g of protein. She takes 42 pills per day, breaking them up into 3 doses of 14 pills.She still prefers it to the liquid formula that always left her feeling full and her stomach sloshy. She hasn't had any issues with taking the pills even though it is a large quantity. She doesn't notice much taste and doesn't have any issues swallowing them. The tablet is coated to make it easier to go down. She generally just drinks a water bottle with them and swallows 2 at a time.

For now Hailey still drinks 10oz with 100g of Ketonex-2 each morning. She used to drink this amount about 3x a day with 215g of powder. Formula will be phased out all together at some point with her on the pills now. Her team didn't want her to quit the formula cold turkey until they were sure that she would respond well to the tablets. The only time since the switch that she had to go back to the 32oz of formula is when she had COVID in July and did get quite sick. We switched her back to Ketonex during this time per her team's suggestion. This allowed us to do continuous feeds every hour to help prevent metabolic crisis at all costs. We were successful and when she started eating her required daily protein again, they allowed her to go back to the tablets.

Even having to go back to the formula for that time Hailey is very happy with her MSUD Easy Tabs. The ease of travel, not having to measure daily, and not having to store her supply with big cans, has made her life much easier. Hailey's metabolic team converted her nutrition needs and prescribed her a number of tablets per day that provided the same values as the Ketonex. Her calories and protein from food has not needed to be adjusted with the switch. She does still take isoleucine and valine but a local compounding pharmacy makes them into capsules for her, that she has been having for several years.



We are very grateful with the tablets and with the company that makes them, Galen. The representative from Galen has been incredibly supportive and most helpful in navigating insurance to ensure that we are not responsible for astronomical copays. This was a great relief since our insurance was fully covering the Ketonex. Hailey has been very happy with her decision to switch to the MSUD Easy Tabs. ■

"FOR THE COMMUNITY, BY THE COMMUNITY" THE CLINIC'S ANNUAL BENEFIT AUCTIONS

The Clinic for Special Children

On select Saturdays in the summer, auction venues in central Pennsylvania, Ohio, and Missouri are abuzz with the rapid fire announcing of auctioneers, greetings of friends, pitter-patter of running children, and the clip-clop sound of horse and buggies arriving. These annual benefit auction days are all in support of a single mission to raise funds to support children and adults with rare genetic disorders at the Clinic for Special Children.

The annual benefit auctions provide vital fundraising support for the Clinic for Special Children. The auctions, along with other fundraisers, contributions, and grants, make up 49% of the Clinic's annual operating budget. In 2019, the auctions cumulatively raised over $1 million – an amazing testament to the power of community. The six annual benefit auctions are held in Union County, PA, Lancaster County, PA, Shippensburg, PA, Shiloh, OH, Memphis, MO, and Blair County, PA. The Lancaster County benefit auction is the longest-running auction for over 30 years!

Local committees of Plain community members, many of whom have family members served by the Clinic, organize the auctions. Starting early in the year, the committees work tirelessly to solicit donations, promote the auction, organize logistics, recruit volunteers, and organize the auction day. Each annual benefit auction is quite literally an event by the Plain community, for the Plain community.

We invite you to join us in 2022 for a day of fellowship and fun at one of our benefit auctions. A tentative schedule of 2022 Clinic for Special Children benefit auctions is included below. For more information on our auctions, please visit www.ClinicAuctions.org or www.ClinicforSpecialChildren.org.

2022 Clinic for Special Children Benefit Auction Schedule

Saturday, June 4 – Union County, PA
Saturday, June 18 – Lancaster County, PA
Saturday, June 25 – Shippensburg, PA
Saturday, July 9 – Shiloh, OH
Saturday, August 20 – Memphis, MO
Saturday, September 10 – Blair County, PA ■

PKU CAMP

By Susan Mays

PKU Camp is a family camp which typically meets annually and provides support, networking, and community engagement to Northwest families and individuals with PKU and Allied Disorders.

There are a plethora of activities to engage in with your families including swimming in the massive pool, sliding down the water slides, a ropes course, miniature golf, cooking demonstrations, and many others. The in-person version takes place in Central Oregon near the town of Antelope.

This year PKU camp went virtual for the first time. We had over 500 attendees from 5 different countries and 41 US states. All attendees who registered in time received a box which included low-protein food samples, a tee-shirt, a bandana (to help represent your team for the Olympic field games), a craft kit, and a s'mores kit from Cook for Love (cookforlove.org). We tried to capture the very best parts of camp and deliver them through a virtual experience. We had a low-protein cooking demonstration by Brenda from Cook For Love, many breakout sessions covering topics including everything from the early years, entering school, and 504's to young adults and maternal health. Other activities included a book reading by an author with PKU and Olympic games, but most importantly we connected with one another.

This is by far the best thing about camp! There is a sense of community in being surrounded by other families who understand what it is like being on a low protein diet and drinking formula. We started going to camp when our daughter Indigo was 1 year old and we fell in love with it. Since then, we have gone every year and have gotten more plugged in with my husband Adrian serving on the board and me helping out.

When asked, Indigo says one of her favorite things about camp is the food. She is excited that she can have anything on the low-protein buffet line. It always gives her the opportunity to try things she never has before and we always leave with a few new favorites to incorporate at home.

Although it is called PKU camp and was founded by families impacted by PKU, the camp seeks to include all allied disorders. We've had families who live with a variety of other Inborn Errors of Metabolism including Homocystinuria, Tyrosinemia, Glutaric Acidemia and Maple Syrup Urine Disease.

We hope to return to an in-person camp again next year with dates reserved from June 2-5, 2022. ■

CALLING ALL NEW ENGLANDERS

By Susan Neeldeman

Do you live in the United States of America and reside in the states of Massachusetts, Connecticut, Rhode Island, Vermont, New Hampshire, or Maine? The New England Connection for PKU and Allied Disorders (NECPAD) wants to hear from you! NECPAD is a non-profit organization that supports MSUD Families in these states. NECPAD offers support, social events, financial assistance, scholarship opportunities, and advocacy for those living in New England states. If you would like to learn more about the group, join the NECPAD E-mail list, come to an event, ask for financial assistance, or even just say, "hi", visit them online or email them at necpad.org@gmail.com

Website: necpad.org Facebook: @necpad Instagram: @necpadorg

RESEARCH

METABOLIC CONTROL AND 'IDEAL' OUTCOMES IN LIVER TRANSPLANTATION FOR MAPLE SYRUP URINE DISEASE

JOURNAL OF PEDIATRICS 2021

Summarized by Dr. James E. Squires, UPMC Children's Hospital of Pittsburgh

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Maple syrup urine disease (MSUD) is a challenging disorder where the body is unable to process specific branched-chain amino acids because of dysfunction in the branched-chain α-ketoacid dehydrogenase (BCKD). Under normal circumstances, the protein that a person consumes is broken down into amino acid subunits which are then further processed by BCKD to enable essential functions of the body to occur. In MSUD, the inability to process certain amino acids (the branched-chain amino acids) results in a build up of potentially toxic by-products which can be extremely harmful, even resulting in death. Non-surgical treatments consist primarily of dietary restriction of protein and monitoring bloodwork for increases in toxic by-products that can serve as a warning for more serious complications. However, even under the best circumstances, complications such as intellectual disability and psychiatric illness are common.

Liver transplant has been used to treat MSUD by providing enough of the missing BCKD to enable the body to process the branched-chain amino acids more easily, thus preventing buildup of the toxic substances that drive morbidity. Previous reports have documented short term improvement; however, data on the long-term safety and efficacy of liver transplant has been lacking. Additionally, we recognize within the liver transplant community that our historical benchmarks of success – patient and graft survival – does not fully capture the complexity of transplant related outcomes as it relates to complications that can be seen from the immunosuppressive medicines that are needed after transplant. In order to address this gap, the transplant community has begun to look at other measures that reflect a successful outcome following transplant, including the design of an 'ideal' outcome composite that looks not only at the transplant recipient and their graft organ, but also at other factors that can at times complicate the health and well-being of a transplant recipient including kidney function, the development of infections, and need for complex treatment regimens that require many different medicines.

In our report, we looked at the largest number of patients with MSUD who received a liver transplant (number=77). We found that the transplanted liver was able to provide enough BCKD function, even in patients over 10 years removed from their transplant, to enable processing of the branched-chain amino acids and prevent dangerous increases in the toxic byproducts, even with diets that are unrestricted. 100% of the transplant patients were alive, no patients had a metabolic crisis, and only 1 patient required a second transplant. Still, when applying the ideal outcome metric to the MSUD transplant recipients, we found that only 39% achieved optimal post-transplant status. Thus, while long-term outcomes in liver transplant recipients for MSUD are promising, continued efforts are needed to optimize metrics that can better assess ideal health and well-being post-transplant. ■

MSUD FAMILY SUPPORT GROUP SCIENTIFIC ADVISORY BOARD MEETS

By Karen Dolins, Research Lead

Our Scientific Advisory Board (SAB) met remotely in June to discuss advances and directions for future efforts. MSUD FSG board members Sandy Bulcher (President), Ed Fischler (Vice-President) and Karen Dolins (Research Lead and Secretary) were in attendance.
Top on the list of items discussed was our MSUD Registry (see article and ad in this newsletter). Increasing participation is essential. Suggestions for promoting enrollment were discussed.

Other projects discussed include:

✔ Adult neuropsychology study – this project is now recruiting (see article in this newsletter).

✔ MSUD Cow project – the birth of a calf with Classic MSUD calf is expected in September and will be used to study gene therapy.

✔ Home leucine monitor – a lab at Columbia University is working on technology which, if successful, would be the first step in developing a monitor which would allow the testing of blood leucine levels in the home.

✔ Messenger RNA – preliminary research is being conducted in mice by Moderna to determine whether therapies using this technology are feasible for MSUD.

The SAB agreed that it would be worthwhile convening a Scientific Meeting on MSUD, bringing researchers together to present on what we know and discuss suggestions for future research. Plans are underway to convene next summer before our Symposium. ■

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

By Dr. Jessica Gold

We continue recruiting for our study at the Children's Hospital of Philadelphia on Neurocognitive Outcomes and Quality of Life for Adults with MSUD. Currently, 10 participants are enrolled. Overall, our goal is to enroll 25 adults with MSUD and 25 unaffected siblings or acquaintances. Online surveys on decision-making, life skills, and quality of life have been distributed and we are excited to see the first set of results.

Importantly, due to the generosity of the MSUD Family Support Group through money raised with the Million Dollar Bike Ride, we are able to offer financial support for travel to complete in-person intellect and executive function testing. If you are between 21-35 years-old and may be interested in participating in this study or have any questions, please contact Jessica Gold at goldj@chop.edu ■

GENE THERAPY STUDY SUPPORTED BY MILLION DOLLAR BIKE RIDE FUNDS PUBLISHED

Hot off the press! Dr. Jenny Greig's work on liver and muscle-directed gene therapy has been electronically published ahead of print in the prestigious journal Molecular Genetics and Metabolism.

Muscle-directed AAV gene therapy rescues the maple syrup urine disease phenotype in a mouse model. 2021 Aug 17;S1096-7192(21)00767-8. doi: 10.1016/j.ymgme.2021.08.003.

You can learn more about Dr. Greig's work by reading her article in the Fall 2020 issue of this newsletter.

MSUD IN EGYPT

By Rofaida M.Magdy
Metabolic and Genetic Unit, Department of Pediatrics, Faculty of Medicine, Sohag University, Sohag, Egypt

MSUD (Maple Syrup Urine Disease) is an amino acid metabolism disorder resulting in accumulation of the branched chain amino acids (BCAAs) leucine, isoleucine, and valine. It has an autosomal recessive pattern of inheritance that runs in families and so parental consanguinity (descended from the same ancestor) increases risk (1). High rates of consanguineous marriage among Egyptian youth (13 – 35 years) has been reported in Egypt (27.4%). It is highest in rural upper Egypt (43.6%) and lowest in urban lower Egypt (13.2%) (2).

In Egypt, there was no nationwide newborn screening targeting Inborn Errors of Metabolism (IEMs). In November 2015, the disability reduction authority in the Ministry of Health established a national screening program for phenylketonuria (PKU). MSUD cases are diagnosed either by selective screening for symptomatic cases or by high-risk screening for family members with an index case of MSUD or previous sibling deaths of unknown cause.

Diagnosis mainly depends on tandem mass spectrometry analysis showing highly elevated branched chain amino acids and ratios: leucine:isoleucine serum levels (normal up to 270 µmol/L), leucine:alanine ratio (normal up to 2.28), leucine:phenylalanine ratio (normal up to 8), Valine serum level (normal up to 198 µmol/L). Only a few cases confirmed with molecular diagnosis to enable prenatal diagnosis in future pregnancies.

There is usually a delay between initial symptoms and the confirming diagnosis. This delay in diagnosis can be caused by: (1) the clinical manifestations at initial assessment which resemble neonatal sepsis; (2) delay in referral from primary units, (3) the limited number of tandem mass spectrometry resulting in the need for samples to be transported over distance and a delay in obtaining results.

After diagnosis, specific management is started immediately using a special formula for MSUD. Special formula is available free in multiple genetic units all over our country. All MSUD patients identified and receiving special formula are registered with the disability reduction authority in the Ministry of Health.

There are 230 MSUD patients registered in Egypt. We believe that this number is much lower than the actual number of MSUD patients as many die before diagnosis. Also, sampling from patients on total parenteral nutrition using amino acids gives unclear results and there is a lack of experience and awareness about IEMs in some NICUs.

It is evident that presymptomatic diagnosis of MSUD is essential for better outcomes, but at this time patients diagnosed through high-risk screening are a minority in Egyptian MSUD patients.

References:

(1) Mohamed MM, Bakheet MA, Magdy RM et al. The clinico-radiological findings of MSUD in a group of Egyptian children: Contribution to early diagnosis and outcome. Mol Genet Genomic Med. 2021;00:e1790.
(2) "Consanguineous Marriage among Egyptian Youth: Secondary Analysis of Survey of Young People in Egypt, 2014". The Egyptian Journal of Community Medicine.2017; 35 (2):85-94. ■

WANTED: MSUD SPEAKERS

VMP Genetics has developed a Patient-Teacher Registry to help teach physicians and health professionals about amino acid disorders. Patients and/or family members who are interested in telling their stories in a medical classroom setting can register to be placed in a database of speakers. If medical educators are looking to include the patient story in a teaching session, the Registry will connect the educators and the "patient teachers."

If you or a loved one are interested, VMP Genetics is always accepting new speakers! If interested, please complete the registration form and consent form (https://www.vmpgenetics.com/edu-services/patient-teacher) and submit them to Jacob Athoe at PatientTeacherRegistry@vmpgenetics.com

Please help VMP Genetics in their efforts to raise awareness about amino acid disorders through this innovative educational outreach to the medical and health care communities. For more information about this project, please contact Jacob Athoe at PatientTeacherRegistry@vmpgenetics.com ■

WHAT IS THE MOST IMPORTANT THING YOU CAN DO TODAY TO HELP IMPROVE THE LIVES OF THOSE WITH MSUD?

Participate in the MSUD REGISTRY!!!

WHAT IS IT?

Our patient registry collects information about people affected by MSUD. Our registry is hosted by CoRDS (Coordination of Rare Diseases at Sanford).

WHY JOIN?

A registry allows clinicians and scientists to learn about how MSUD affects those with this condition and their families. They use this information to plan research, develop therapies, and hunt for a cure.

WHO SHOULD COMPLETE THE REGISTRY?

- Individuals with MSUD or a family member regardless of MSUD type.
- Individuals with MSUD who have received a liver transplant.
- Family members of a deceased person with MSUD.
- Anyone with MSUD from any country in the world.

WHAT IF I DON'T SPEAK ENGLISH?

Our registry questionnaires are available in Spanish.

HOW DO I REGISTER?

The registry is available through our website: www.msud-support.org or https://research.sanfordhealth.org/rare-disease-registry
Need a hard copy or Spanish questionnaire? Call CoRDS at 877-658-9192 or email them at cords@sanfordhealth.org.

HOW LONG WILL IT TAKE TO COMPLETE?

Completing the questionnaires will take approximately 30-40 minutes. It does not need to be completed all at once.

CONCERNED ABOUT SECURITY?

Your information is secure. Personally identifiable information will not be shared with researchers.

QUESTIONS? CONTACT KAREN DOLINS AT KAREN.DOLINS@GMAIL.COM OR 914.391.2982.

ORGANIZATIONAL AND PROFESSIONAL CONTACTS

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

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