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MSUD NEWSLETTER

Published by the MSUD Family Support Group ■ Volume 37-2 ■ Fall 2019



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

By Ed Fischler, VP MSUD Family Support Group

The MSUD Family Support Group has 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. In support of its mission, the ODC sponsors and promotes the annual MDBR. The funds raised are used to support pilot research grants for the rare disease represented by each participating team. This year's event was held on the streets of Philadelphia on June 8th.

Representing the MSUD Family Support Group (and shown in the photo) were: Taryn Kessel of Chicago with her husband Doug; Karen, Jerry and Jessica Dolins of New York with Hannah; and Herb Foster of Boston with his brother-in-law Steve. Also, we had the support of other families who organized fund raising events and efforts in their home towns. These included the families of Jordan Coleman, Andrea Alba, Michelle Flanagan, Mindi Penner, Susan Mays, Wayne McGill and Chad Farquharson, and Edward Fischler.

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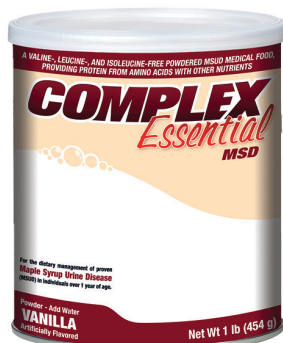
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The information contained herein does not necessarily represent the opinions of the MSUD Board, Medical or Nutritional Advisors, or all of our members. Before applying any of the information contained in this newsletter, you must consult a MSUD specialist.

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MSUD FAMILY SUPPORT GROUP NEWS

FROM THE PRESIDENTS' DESK



By Sandy Bulcher

The MSUD Family Support Group Board of Directors meets annually in addition to holding quarterly conference calls. This allows us to make continued progress towards our goals. Our face to face meeting was held the last weekend of June at the

beautiful home of board member Herb Foster and his wife Diane in Falmouth, MA. Thanks Herb and Diane, for being such welcoming and gracious hosts.

During our 2 day meeting, we discussed our goals of connecting and engaging MSUD families, progressing research to improve the lives of those with MSUD, and advocating to meet the needs of our community, and identified action steps needed to accomplish these objectives. We also worked on our budget which keeps us fiscally responsible.

Research remains a major focus. Researchers are interested in the worldwide prevalence of MSUD, and knowing where those affected are located. We have begun to research how we can best gather this information. We also continue to explore other treatment options.

In the past 6 months, several board members attended professional meetings. Karen Dolins attended the National Organization of Rare Diseases meeting in Washington DC, and Jordann Coleman traveled to DC for Hill Day to promote the Medical Nutrition Equity Act. Karen Dolins attended the Rare Drug Development Symposium in Philadelphia prior to participating in the Million Dollar Bike Ride with fellow board member Herb Foster and others. Karen Dolins and I collaborated with the University of Pennsylvania's Orphan Disease Center who hosted an MSUD Think Tank meeting in Philadelphia. Read more about these events in this newsletter.

'Presidents' Desk' continued on page 5



EDITOR'S NOTE

By Dr. Karen Reznik Dolins, EdD, RDN, CSSD

As you can see from the contents of this newsletter, our support group has been busy! Our life events section highlights the rewarding lives many are living. Thanks

to Andrea Alba for sharing the story of her niece and nephew who apparently lived with undiagnosed MSUD until their 30's.

The board has been actively attending to fundraising (Million Dollar Bike Ride), advocacy (Hill Day for Medical Nutrition Equity Act), and research efforts (Think Tank). You can read all about these efforts in this issue.

Also in this issue you'll find advice on vaccinations and isoleucine and valine supplementation from members of our advisory board, and can learn about a cow model for MSUD and what it has to offer.

We thank our valued advertisers who keep us advised of new and improved products essential to MSUD.

I hope you will find this issue of the MSUD Family Support Group Newsletter to be uplifting and motivational. We've come so far. With each and every one of us pitching in, there's no limit to what we can accomplish!

Please contact me at Karen.dolins@yahoo.com with suggestions for articles for our Spring 2020 newsletter ■

MSUD AND IMMUNIZATIONS

By Jessica Scott Schwoerer, M.D.

I have a checklist that I review when seeing patients with Maple Syrup Urine Disease (MSUD) or other metabolic disorders.

- Growth
- Development
- Sick day diet
- Immunizations

Illness prevention is key to the care of a patient with MSUD. Illness brings the misery of sickness, the stress of pushing a sick diet, overnight feeds, stat amino acid level(s) and the possibility of hospitalization. With every illness, there is the risk of metabolic decompensation and risk of brain injury and death. Each illness we can prevent is important to the patient and their family.

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Immunizations are an important part of illness prevention. Immunizations can prevent or lessen the severity of illness. This leads to fewer episodes of illness and all that accompany it - including the risk of a severe decompensation. For all patients, including those with MSUD, the full routine immunization schedule is recommended to prevent common and potentially severe illnesses. As we head into the fall months, it will be time for the annual Influenza immunization too. Make sure to schedule the "flu shot" for all family members to decrease the risk for influenza in your household.

When I talk with families, the most common reason for not immunizing is concerns about safety. There is a lot of information in the news, internet and social media about immunizations and their safety. It is a lot to wade through. The bottom line – immunizations are safe. Adverse reactions are rare. Years of research have shown that immunizations, including the MMR (Measles, mumps, and rubella) vaccine do not cause autism spectrum disorders. The research study that connected immunizations and autism over 20 years ago was wrong.

So, when you are at your physician's office, make sure you and your child are up to date on immunizations. And put getting the influenza immunization at the top of your fall to do list.

WEBSITES FOR MORE INFORMATION:

Immunization schedule - <https://www.cdc.gov/vaccines/schedules/>

Immunization safety information - <https://www.cdc.gov/vaccines/parents/vaccine-decision/index.html>

***Editor's Note:** Check with your metabolic clinician about the need for dietary changes when receiving an immunization ■

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'Presidents' Desk' continued from page 3

For us to reach our goals it will take both financial and personnel resources. Please consider fundraising, donating, and volunteering. A donation slip is included in this mailing. We appreciate your generous financial support which helps cover expenses associated with our efforts, including our biannual symposium.

MSUD Symposium 2020 will be held in Columbus Ohio at the Embassy Suites Airport June 25th-27th. Start planning now to attend the fun and educational 3 day event. Hope to see you there!

I am joined on the board by:

Vice President Ed Fischler -FL,

Treasurer Dave Bulcher -Ohio,
Secretary Karen Dolins- New York
and members Herb Foster - MA, Ivan
Martin- PA and Jordann Coleman - CA.

As always, feel free to contact me or any board member if you have any questions, concerns, or comments. My contact information is 740-972-5619, sandybulcher@gmail.com. ■



MSUD Board from left: Herb Foster, Jordann Coleman, Ed Fischler, Karen Dolins, Sandy Bulcher, Dave Bulcher, Ivan Martin

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'Bike Ride' continued from front cover

Let's thank the whole team who raised 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 those affected by MSUD. These families have done some of the heavy lifting that will be necessary to fund the research necessary to achieve these goals. This year, a total of 466 donations for over \$65,000 was contributed to the MDBR campaign. The University of Pennsylvania/Orphan Disease Center will match our amount with an additional \$30,000, boosting the total amount raised to over \$95,000!

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 seeks better treatments for MSUD. The other will be directed towards finding a cure for MSUD. The donations

and matching funds are to be evenly split between the two projects. The final project selection will wrap up by November so that projects may begin early in 2020.

This was the second year of our participation in the MDBR. In 2018 the donations raised enabled a gene therapy research project that is currently underway at the Penn Medicine / Orphan Disease Center. The project will conclude later this year. Hopefully, we'll be invited back to participate in the 2020 MDBR.

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



FAMILY NEWS

A SPECIAL DEDICATION

By Susan Needleman, *Classic MSUD Adult Age 29, New England Connection for PKU and Allied Disorders Outreach & Allied Disorders Coordinator*

On May 4, 2019, the New England Connection for PKU and Allied Disorders (NECPAD) held their 6th Annual 5K Race, in Brighton, MA. This year the race was dedicated to Scott Foster and his family, who have given so much to the MSUD community.

Scott C. Foster was born to Herb and Diane Foster, on August 17, 1971, and quickly made history as the first baby to be diagnosed with MSUD through newborn screening testing in the State of Massachusetts.

At the age of 22, Scott, came down with the flu, which caused a rapid metabolic decompensation and brain edema. Sadly Scott lost his life on May 9, 1994.

For the next ten years, his family held fundraisers in Scott's name, including ten dinners, road races, and golf tournaments.

In recent years the Foster Family made history again, as their third child and second with MSUD, Katie, had two healthy children, Kayla and Pete. With their births, Katie opened the door to show how MSUD women could safely go through a pregnancy and have their own kids.

In the past year the Foster family has graciously donated half of the money they raised over the years to NECPAD and half to the MSUD Family Support Group, totaling \$35,188 each. NECPAD will spend their share of the money

to help families with MSUD and Allied Disorders in New England (Massachusetts, Connecticut, Rhode Island, Vermont, New Hampshire, and Maine. NECPAD would like to thank the Foster Family for their generous donation. ■



HATS OFF TO THE GRADUATE

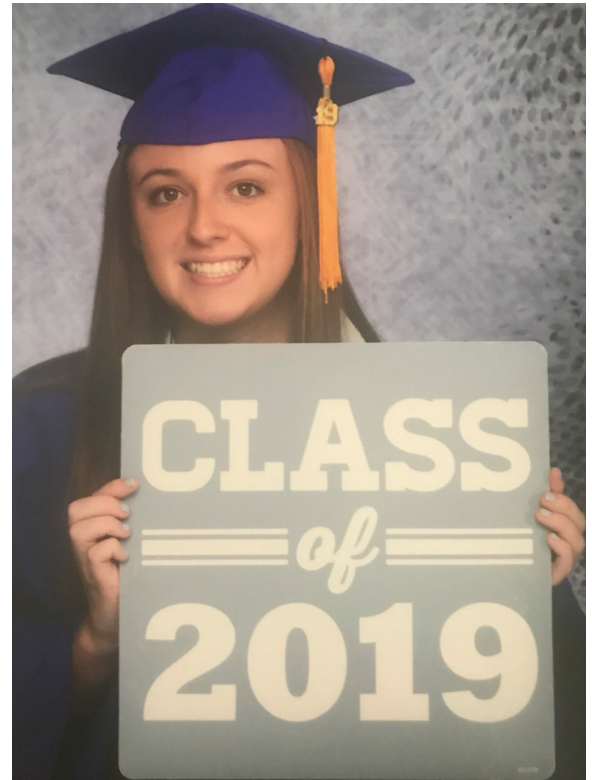
By Michelle Flanagan

The first day of kindergarten seems like it was just yesterday. I was a ball of nerves as I left my medically fragile little one in the care of a school that had no experience with MSUD. Over time we fell into a routine and it didn't seem so scary. Hailey's health was always taken seriously and I'm happy to say that the school system was quite supportive no matter what she was going through.

In what seems like the blink of an eye, my pigtailed kindergartener has graduated high school. She graduated as an honor roll student. She received the lottery funded HOPE scholarship that Georgia provides to students who maintain a B average through high school. HOPE covers tuition and fees as long as she attends an in-state

university and meets GPA requirements. We moved her into her college dorm this weekend. She is attending the University of West Georgia, where she plans to pursue a Bachelor of Science in Nursing. We have an altered meal plan, emergency medical plan with health services, student accommodation plan for housing, and a school assigned Student Health Advocate that will keep Hailey on track during college despite living with a serious medical condition.

I couldn't be more proud of her for going after what she wants in life. I admire her drive, courage, and determination more than she could



possibly know. Congratulations to my 2019 high school graduate and official college freshman! ■



COLLIN BURLESON

CLASSIC MSUD, AGE 19

— **By Mandie Burleson, Mom**

Collin was born on July 28, 2000 and at that time there was no newborn screening in Arkansas where we lived. Collin was not diagnosed until he was 8 days old and in a very critical state with a blood leucine level over 3000. He was comatose, intubated, required dialysis, and had to have a g-tube placed.

After diagnosis we were told that Collin more than likely would have some developmental delays due to late diagnosis and the cerebral edema that occurred with such high leucine. Collin had struggles early on and at 9 years old also received a secondary diagnosis of Autism. He is very high functioning and very bright, however he still struggles with some things.

Collin's goal was to graduate high

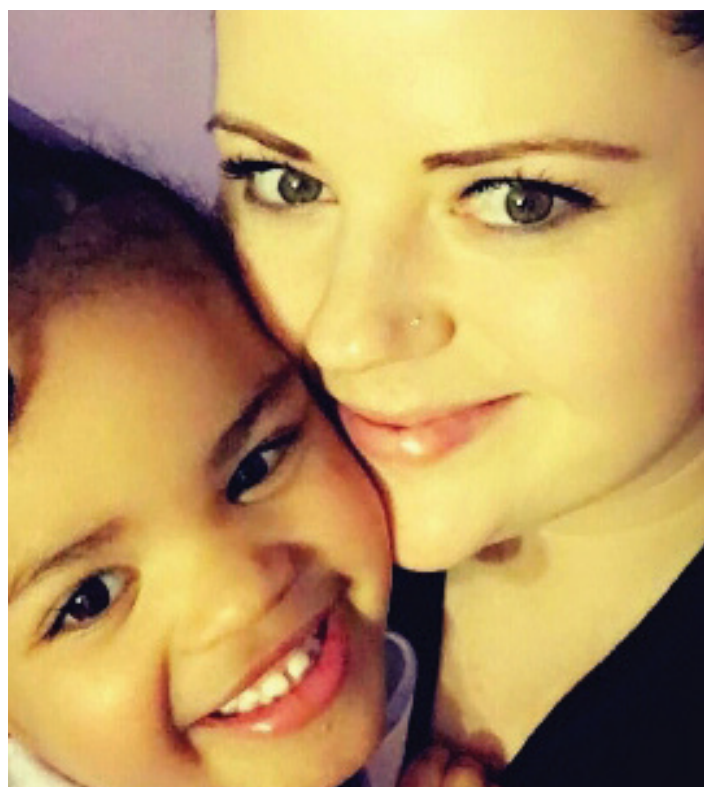
ANDREA OBULATA- EZELECHE

AUGUST 18, 1987 – JUNE 22, 2018

By Andrea Alba

Last June 2018 I brought my beautiful sweet niece Andrea, age 30, to Sainte Boniface Hospital in Winnipeg Canada. What began with a gastrointestinal norovirus had progressed to episodes of mental confusion. As she began having seizures the ICU doctors desperately tried to save her life, but failed when she suddenly and tragically lapsed into a coma and died, leaving behind a 3 year old daughter.

Andrea was healthy her whole life, and the doctors didn't know why she became deathly ill. The cause of death was listed as suspected encephalitis of unknown causes.



Several months after her death her older brother, also in his 30s, experienced the same illness. This time the doctors performed exhaustive testing and diagnosed MSUD. Post-mortem gene sequencing revealed that my niece also had MSUD. As far as we know, they are the first individuals to be diagnosed with this illness in adulthood.

school and receive a regular diploma just as his peers did. Collin had an IEP in school which provided him with the resources and services needed to make this happen. He really enjoyed his high school years despite many illnesses, hospitalizations, missed classes and ALOT of extra work. Collin was voted PROM KING by his class and walked across the stage during the graduation ceremony VERY proud of his accomplishments.

Collin also makes sure to spread awareness about MSUD to everyone he meets, and has presented to the entire school district as well as community on

Rare Disease Awareness Day for the past 4 years.

Collin has big plans for the future. He is working now on finding a good career path and plans to open his own business in our small community. He also plans to become an author, and has a strong interest in writing a children's book about living with MSUD, possibly even teaming up with his siblings to write a story about what it is like to have a sibling with MSUD.

One thing is for certain: whatever path he decides to take, he will succeed and he will not let MSUD or Autism get in his way. ■

We could not save our niece, but are committed to doing everything possible to help our nephew and others living with this disease. This is why I decided to participate in the Million Dollar Bike Ride to raise funds for MSUD research. I was joined by my sister whose husband owns a bike shop in Thunder Bay, Ontario. They hosted a BBQ and bike ride. All donations from these events went to my donor page.

We are so happy to be part of a bigger "family" looking into research for improved treatments of this disease.

Attached is a picture of my niece Andrea with her little daughter Maya age 3 who lost her mom to MSUD. ■

DAVID FISCHLER | CLASSIC MSUD AGE 31

David Fischler, the son of Edward and Lynn Fischler was born in 1988 in Atlanta, GA. David has an older sister and twin brother, both unaffected by MSUD. Within days of his birth, David was showing signs of severe stress and illness. He was diagnosed with MSUD at 16 days old and spent two months in the intensive care unit at Egleston Children's Hospital, Emory University in Atlanta. His first several years were very difficult, requiring frequent visits to Egleston. Fortunately, David's doctors had experience with MSUD and helped him to recover from the several crises he experienced in his early years.

As he got older and learned with his parents how to manage MSUD, he became an excellent student and was able to participate in school activities and sports. He played "Tee" baseball, became the starting goal keeper on his soccer team, and was a member of his high school marching band. After graduating from high school, he earned admission to the Georgia Institute of Technology (Georgia Tech) in Atlanta. Despite hospitalizations resulting from MSUD complications, he was an outstanding student and a

'David' continued on page 13



MSUD BOWLING EVENT



Front (left to right): Josiah Velasquez, Jeramiah Velasquez; back (left to right): Leanna Peters, Susan Needleman, Jaden Flores, Mellissa Berman

— **By Susan Needleman**
29 Year Old Classic MSUD Adult

New England Connection for PKU and Allied Disorders Outreach & Allied Disorder Coordinator

Massachusetts and Connecticut MSUD Families gathered together on August 18 for an afternoon of bowling. Six individuals with MSUD, ranging in age from 3-37, and their families, for a total of 23 people, enjoyed an afternoon of bowling, meeting others with MSUD, and eating delicious low-protein foods, donated by Ajinomoto Cambrooke.

The event was sponsored by the New England Connection

'Bowling' continued on page 11

'Bowling' continued from page 10

for PKU and Allied Disorders (NECPAD), which supports families with MSUD and allied disorders in Massachusetts, Rhode Island, Connecticut, Vermont, New Hampshire, and Maine. NECPAD supports these families, assisting them with medical expenses, social events, advocacy, research and awareness.

To find out more information, contact NECPAD at:
New England Connection for PKU and Allied Disorders

Email: necpad.org@gmail.com

Web: necpad.org

Facebook: <https://www.facebook.com/necpad/>

Instagram: [necpadorg](https://www.instagram.com/necpadorg)

Direct Email: slneedleman@yahoo.com

Phone: 781-420-2676 ■

ADVOCACY

2019 MEDICAL NUTRITION EQUITY ACT HILL DAY

By: Jordann Coleman, Board Member and Advocacy Chair



On May 7, 2019 I traveled to Capitol Hill as a representative of the MSUD Family Support Group to lobby our members of Congress and encourage them to support the Medical Nutrition Equity Act (H.R. 2501). I was one of about 100 advocates participating in the Hill Day coordinated by the Patients & Providers for Medical Nutrition Equity. This coalition is made up of nearly 40 medical professional & disease organizations who rely on medical nutrition.

Prior to the Hill Day, a training session was held for all of the advocates. We were given information on the scope of the legislation, the bill number as it had just been introduced in the House of Representatives the week prior, pointers on how to share our stories, and how to ask the Congressional Members to co-sponsor the legislation. We sat at tables with other advocates from our states and were able to practice our stories and our "ask" (what we are asking the Members of Congress to do).

The next morning, the group got together again for a brief review and we received our schedules. I was with one other Californian and our schedule consisted of 8 meetings and 5 drop-offs. The coalition scheduled our meetings, so all we had to do was show-up and tell our

'Hill Day' Continued on page 12

'Hill Day' Continued from previous page

stories. Our first meeting was with my Congressman's health legislative assistant. On Hill Days, it is rare that you will meet with elected officials. Meetings are typically held with staffers who are usually in their 20s & early-30s. I brought a picture of my son, Carter, and shared our experience in obtaining medical foods & formula, why they are so important to his overall health, and my concerns for his future if this legislation is not passed. My advocate partner shared her story and after answering a few questions, our meeting was over after about 15 minutes. Our subsequent meetings all followed this pattern. Overall, the meetings went well and most of the staffers told us they believed their legislator would be in favor of co-sponsoring and supporting the legislation. It was stressed on us in our training meetings how important follow-up is to getting co-sponsorship, so I obtained business cards for all of the staffers we met with and sent emails thanking them for their time a few days later. Several gave me time frames in which to check in on the status of their boss' decision and I spent the next few weeks sending follow-up emails to see if a

decision had been made. I am happy to share that one of my scheduled meetings resulted in a co-sponsorship! I am still following up with the offices who have yet to give me an answer and hope for some additional co-sponsors.

My Hill Day experience was great. However, of the eight meetings I attended, 7 were with Democratic offices (I am from California!). In order for this legislation to pass, bipartisan support is needed. If you are represented by Republican members of Congress, I urge you to contact them to support this bill. All elected officials have local offices so you won't need to travel to Washington DC to make your voice heard. I am happy to help you prepare for a meeting with your Congressional office to talk about the Medical Nutrition Equity Act. Please contact me at coleman.jordann@gmail.com.

In addition, we need more stories from those with MSUD to share why Medical Foods are so important. Please visit nutritionequity.org/share-your-story/ and share your story.

As of the publishing of this newsletter, there are 35 co-sponsors to this Bill. ■

STARTING AND MAINTAINING A SUPPORT GROUP

By Sandy Bulcher, President MSUD Family Support Group

On July 19th, I gave a presentation, titled: Starting and Maintaining a Support Group at the SERGG (Southeast Regional Genetics Group) meeting at the Renaissance Hotel in Asheville, NC. SERGG is a network of genetics and newborn screening providers in the Southeast U.S. Their goal is to expand and improve genetic resources and services in their region.

The organizers felt that others could benefit and learn from our success, and I was asked to participate in the consumer breakout. I informed attendees that the MSUD support group unofficially began in 1979 when families were communicating via a circulating letter. The first symposium was held in Goshen, Indiana in 1982 and the first newsletter was mailed to families in 1983. By 1994, our organization was a 501c3 tax exempt organization.

I explained the fundamentals needed to become a 501c3 tax exempt organization, which include:

bylaws, which define how the organization will operate, .

Articles of Incorporation with the state, which define what the organization will do, and IRS paperwork

The process can be quite time consuming and slow.

I also shared what I felt were the most important issues for a support group to address: Communication, Fundraising, Advocacy, and Developing Relationships.

It's important for our organization to be visible and active at meetings throughout the year as it is an essential part of networking and increasing awareness of MSUD. It is a goal of the MSUD Family Support Group board to participate in several meetings outside of MSUD symposiums each year. ■

HELP TEACH ABOUT MSUD



VMP Genetics provides direct patient care, physician support services and education services, and is looking for volunteers to help educate the medical community about Maple Syrup Urine Disease.

FACT! Teaching about metabolic diseases in medical school and residency programs is poor.

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.

THEY NEED YOUR HELP!

VMP Genetics believes 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. VMP Genetics also believes 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.

THEY ARE LOOKING FOR...

Patients and/or family members who are interested in telling their stories in local medical classroom settings... VMP Genetics is 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 they secure funding, VMP Genetics is also 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 that can be downloaded into a lecture about that disease or relevant biochemistry to enhance the learning potential of the session.

Please help raise awareness about Maple Syrup Urine Disease through this innovative educational outreach to the medical community. For more information, please contact: PatientTeacherRegistry@vmpgenetics.com ■

'David' continued from page 10

member of the college marching band. David proudly graduated with honors from Georgia Tech in 2011.

After receiving his undergraduate degree, David began graduate studies in the Department of Chemistry at the University of Georgia. While at UGA, David worked on a doctorate degree in Analytical Chemistry. In May, 2019, he received a Ph.D. degree in Chemistry. He now holds an Oak Ridge Institute for Science and Education

(ORISE) postdoctoral fellowship position located at the Centers for Disease Control (CDC) in Atlanta.

David has already accomplished much and is a very caring and amiable person who goes out of his way to help others. But he is also aware that MSUD is the “wild card” in his--and other’s lives. He wants to join others to improve the lives of those with MSUD and hopefully, someday find a cure for this and other diseases. ■

Healthy, Low Protein Recipes

From Dana White Nutrition

ROASTED TOMATILLO SALSA

MAKES ABOUT 3 CUPS

- 6 large tomatillos (about 1 ½ pounds), quartered
- 1 clove garlic
- ½ medium white onion, chopped
- 1 jalapeno pepper, roughly chopped
- Olive oil
- ¾ cup fresh cilantro
- Juice of ½ lime

Preheat oven to 400 degrees F. Place tomatillos, garlic, onion, and jalapeno on a sheet pan. Drizzle with 2 teaspoons olive oil and season with ¾ teaspoon salt and black pepper to taste. Roast for 20 minutes, until tomatillos are tender, set aside to cool slightly. Place roasted vegetables with cilantro and lime juice in a food processor and pulse until smooth. Serve chilled or at room temperature.

Nutrition info per tablespoon: Calories: 5; Total Fat: 0 g; Saturated Fat: 0 g; Total Carbohydrate: 1 g; Sugars: 1 g; Protein: 0 g; Sodium: 18 mg; Cholesterol: 0 mg; Fiber: 0 g



STRAWBERRY LEMONADE SLUSHIES

MAKES 2 SERVINGS

Pass on those convenience store ice drinks laden with sugar and artificial colors. Beat the heat with this seasonal and naturally beautiful sipper.

- 1 1/2 cups lemonade
- 1/2 cup 100% apple juice
- 1 cup strawberries

Place lemonade, apple juice and strawberries in a blender. Blend well and pour through a fine mesh strainer to remove the seeds. Pour mixture into a ice cream machine and run for about 15 minutes or until mixture is as frosty as desired. Pour into glasses and serve.

Calories: 136; Total Fat: 0 g; Saturated Fat: 0 g; Carbohydrates: 35 g Protein: 0 g; Sodium: 2 mg; Cholesterol: 0 mg; Fiber: 1 g

EASY NUT-FREE LEAFY GREEN PESTO

A combination of bright green vegetables and herbs come together to create an inflammation fighting and nutrient rich powerhouse sauce. Not only do leafy green vegetables like spinach, arugula, and fresh herbs contain vitamins and minerals, they're packed with plant-based compounds that protect cells from damage and inflammation. Make a batch of this vibrant and versatile sauce for pasta, sauces, or grilled meat and vegetables.

Makes 24 servings (1 tablespoon = 1 serving)

INGREDIENTS:

- 1 clove garlic chopped
- 2 cups fresh arugula
- 1 cup fresh spinach
- ½ cup fresh parsley
- ½ cup fresh basil juice
- zest of one lemon
- 1 teaspoon kosher salt
- ½ teaspoon freshly ground black pepper
- 1 cup extra virgin olive oil

PREPARATION

1. Place garlic clove in a food processor and pulse to chop.
2. Place arugula, spinach, parsley, and basil to the food processor, followed by lemon juice and zest.
3. Add salt and black pepper and pulse again.
4. With the machine running, slowly pour in olive oil.
5. Continue to blend until all the oil is incorporated and ingredients are well combined.
6. Scrape down the sides of the bowl with a rubber spatula to make sure all ingredients are incorporated.
7. Transfer the pesto to a bowl; use immediately or store as desired.

Nutrition information per tablespoon: 81 calories 9 grams fat 0g carbohydrate, 0g protein

Recipe courtesy Dana white nutrition and very well.com
<https://www.verywellfit.com/easy-nut-free-leafy-green-pesto-4137166>



Photo: www.goodfreephotos.com

ISOLEUCINE AND VALINE:

WHEN IS SUPPLEMENTATION NEEDED?



Left: Rani H. Singh, PhD, RD, LD, Professor and Director, Genetic Nutrition Research Program, Emory University, Department of Human Genetics; Right: Katie Hobson, MS, RD, LD, MNT4P Post Master's Clinical Research Fellow

Amino acids are building blocks used to make proteins. Leucine, isoleucine, and valine are a type of amino acid called branched-chain amino acids (BCAA). These are essential amino acids, which means they cannot be made by our bodies, but must be consumed as part of our diet. The BCAA, along with other amino acids, play a vital role in growth and development.

While MSUD affects the metabolism of all three BCAA, leucine is the most concerning because when it builds up in the body, it has a toxic effect on the brain. Each patient is prescribed a specific protein or leucine goal to ensure they are receiving adequate amounts of BCAA from food without having a toxic buildup of leucine. Dietary goals for isoleucine and valine are not recommended since most foods have nearly double the amount of leucine. Additionally, high dietary intake of isoleucine and valine do not have the same toxic effect on organs as high levels of leucine.

Patients may have elevated levels of leucine in their blood due to consumption of too much protein from food or from the breakdown of their own muscle. BCAA are

stored in the muscles, so during times of illness, fasting or general inadequate energy intake, muscle is broken down for energy and BCAAs are released into the blood.

While patients with MSUD must limit the amount of BCAA in their diets, it is equally important that adequate amounts be consumed to support the many roles BCAA have in the body. BCAA deficiency can result in anemia, skin damage, diarrhea, and poor growth. The metabolic formula prescribed for MSUD patients contains no BCAA; therefore, with a protein-restricted diet some patients may become deficient in isoleucine and valine.

Patients with MSUD may require supplementation of isoleucine and valine if:

- 1) Plasma isoleucine and valine levels are low indicating a patient is not consuming enough dietary protein. Some patients cannot increase their protein from food because their leucine level would increase as well. According to the Genetic Metabolic Dietitian's International guidelines¹, the following are the recommended treatment ranges for plasma BCAA:

LEUCINE:

- Infants & Children <5 years of age: 75-200 umol/L
- Individuals >5 years of age: 75-300 umol/L

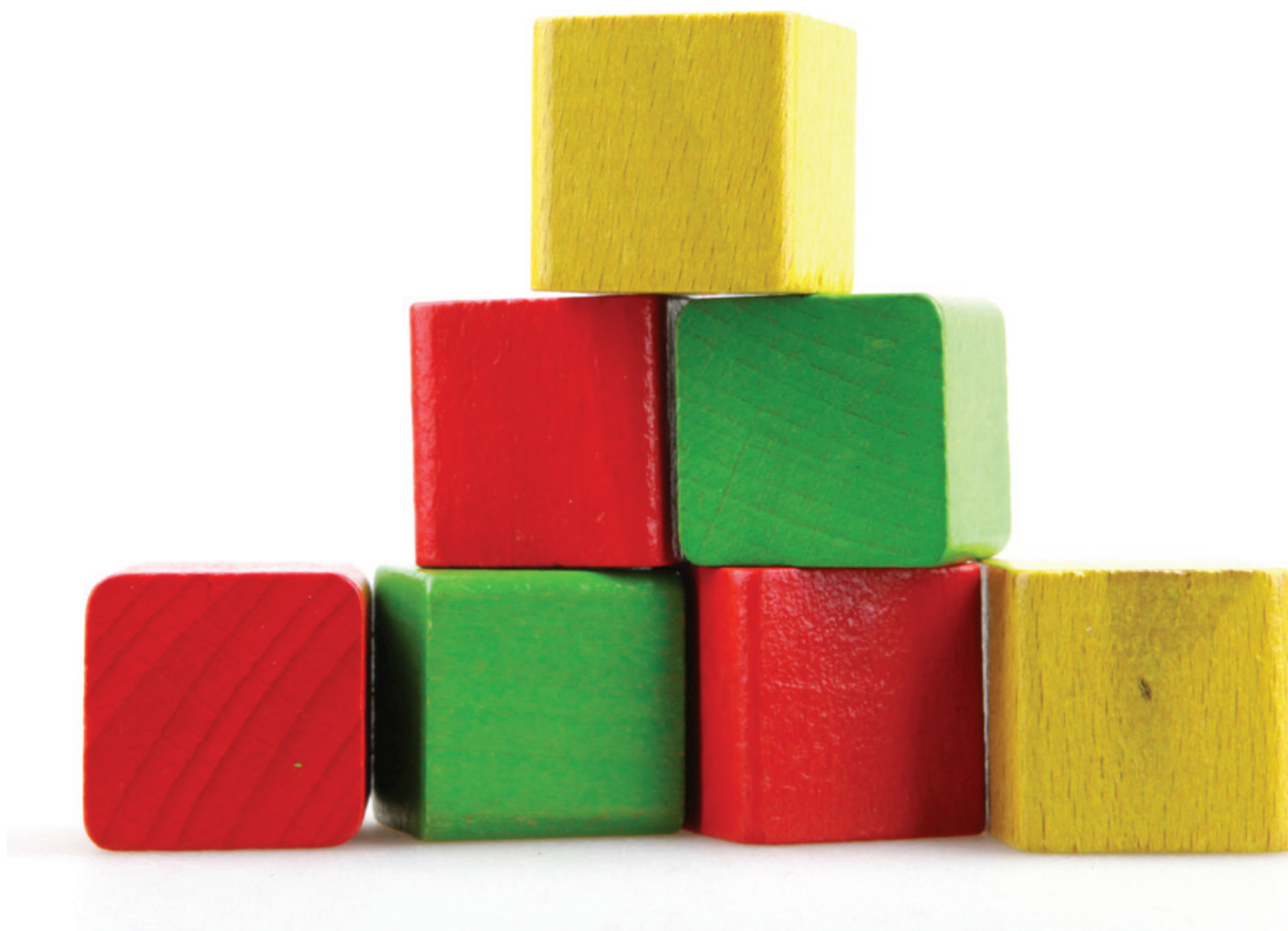
ISOLEUCINE:

- 200- 400 umol/L

VALINE:

- 200 - 400 umol/L

- 2) Leucine levels become extremely high during times of metabolic decompensation. Metabolic decompensation typically occurs when there is inadequate energy intake or illness leading to muscle breakdown. Treatment with isoleucine and valine can



aid in quickly bringing leucine levels down by halting and reversing muscle breakdown. Additionally, isoleucine and valine can have a protective effect on the brain by competing with the transport of leucine at the blood brain barrier. Generous supplementation with isoleucine and valine during decompensations have been shown to reduce the blood leucine concentrations rapidly. Isoleucine and valine supplements have not been associated with any harm, even when the concentrations are higher than the recommended ranges.

supplementation is not always warranted. Some patients with MSUD have higher protein tolerances or formula prescriptions that allow them to consume adequate amounts of isoleucine and valine from food without supplementation. Healthcare providers should assess several parameters before prescribing supplementation such as growth, intact protein intake and protein equivalents from, medical food, and amino acid levels in the blood. ■

¹Nutrition Management Guidelines for Maple Syrup Urine Disease (2013), a collaborative project between Southeastern Region Network (SERN) and Genetic Metabolic Dietitians International (GMDI) see at: www.GMDI.org

Despite the potential benefits of isoleucine and valine,

HOW NOW, RED AND WHITE COW?



Jon Beever, PhD
*Professor and Director,
Genomics Center
for the Advancement of
Agriculture, University
of Tennessee Institute of
Agriculture, Knoxville, TN*

How can cows help with finding a possible treatment for MSUD? Just like humans, cows can also have MSUD. In the Hereford (pictured)

and Shorthorn cattle breeds, mutations are present in the same gene that causes classic or Type 1A MSUD in humans. In Hereford cattle, calves homozygous for a mutation called Q50X have no detectable BCKADH enzyme activity. Consequently, the appearance and progression of the disease in newborn calves

very closely mimics that of severe MSUD cases in human newborns. In contrast to human populations, animals are often selectively bred to reduce the frequency of these mutations in the population, thereby decreasing the incidence of affected calves. Until recently, these mutations, that were discovered almost 30 years ago, were thought to have vanished from the population. However, in the past few years, several cases of MSUD were reported in calves from Indiana and Iowa. Although this was surprising to veterinarians and cattle producers, it has fortuitously provided renewed access to this large animal model for MSUD. Unlike mouse models of MSUD, this model so closely mimics classic MSUD, affected calves may be able to play an important role in evaluating novel treatments such as gene therapy or gene editing. To ensure that MSUD calves can be produced when needed for such research, genetic material from some of these animals has been preserved and is awaiting new therapies to be tested. ■





MSUD RESEARCH

THINK TANK MEETING

By Karen Dolins and Sandy Bulcher

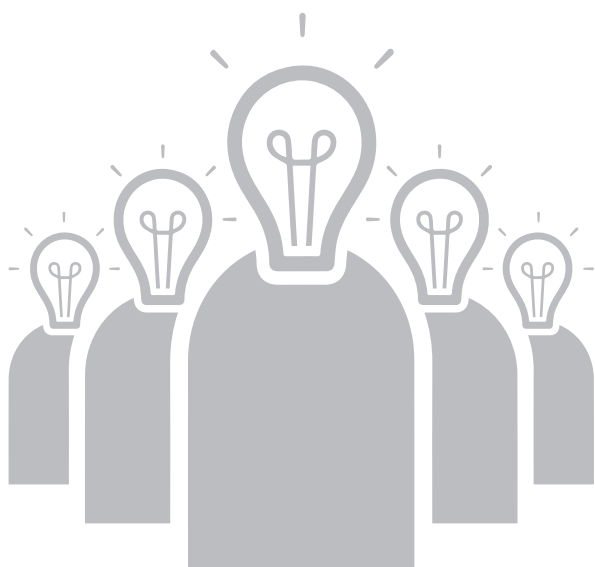
At our last symposium, we were thrilled when Dr. James M. Wilson of the University of Pennsylvania's Orphan Disease Center suggested that we work together to convene a group of researchers and clinicians involved

in MSUD with the aim of identifying gaps in the research and determine priorities for moving forward. This meeting took place on June 20th in Philadelphia, and we represented the MSUD Family Support Group.

Our medical advisor, Dr. Melissa Wasserstein of Montefiore Hospital and Albert Einstein College of Medicine in New York led the meeting. Other attendees included medical advisor Dr. Can Ficicioglu, Children's Hospital of Philadelphia, Dr. Gregory Rice, University of Wisconsin, Dr. Rakesh Sindhi, Hillman Center for Pediatric Transplantation at the University of Pittsburgh, Dr. Jonathan Beever, University of Tennessee, and Dr. Vincent Carson and Karlla Brigatti, Clinic for Special Children. Dr. Wilson was our host, and Dr. Jenny Greig, recipient of the 2018 MSUD Million Dollar Bike Ride grant, was there to present her preliminary research findings on muscle-directed gene therapy.

Another hot topic at the meeting was the possibility of using a type of gene therapy, called messenger RNA

'Think Tank' continued on page 20



'Think Tank' continued from page 19

(mRNA) therapy, as a therapeutic tool. Dr. Greig is investigating the use of mRNA to target liver cells to replace the defective enzyme for metabolism of branched-chain amino acids. There is interest in determining whether mRNA can be used during a metabolic crisis to rapidly reduce leucine levels and protect the brain as well as for ongoing, daily management of MSUD.

When it was our turn to present, we noted some of the challenges to living with MSUD, which include:

- Threat of metabolic decompensation
- Inability to monitor BCAA levels regularly using a home monitor
- Determining the best diet for each individual and improving compliance
- The effect of MSUD on the brain

We noted that researchers have approached us to ask for our support with various projects, but that we'd like to drive research into

areas that we feel are important.

We had a lively discussion about future research priorities, including gene therapy, the need for neuropsychological testing and quality of life studies, the need to continue the work begun at Emory on a patient registry, to work with the community to compile natural history data, and the need to collaborate with researchers and clinicians around the world.

We left the meeting feeling excited about the opportunities for further improvements in MSUD care and potential molecular therapies. ■

UPCOMING EVENTS

2020 MSUD SYMPOSIUM

Sandy Bulcher, MSUD Board President and Symposium Coordinator



The 2020 MSUD Symposium will be held in Columbus, Ohio on June 25-27th at the Embassy Suites by Hilton Columbus Airport, 2886 Airport Drive, Columbus, Ohio, 43219. Attendees will hear presentations on medical and dietary treatment, transplantation, research projects, and advocacy. There will also be ample time for social interaction to visit with old friends and make new ones. Mark your calendars to attend. More information will be available in early 2020. ■