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MSUD NEWSLETTER

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MSUD FAMILY SUPPORT GROUP INVITED BACK TO THE MILLION DOLLAR BIKE RIDE!

By Ed Fishler, Vice President, MSUD Family Support Group

Our community needs improved treatments and hopes for a cure for MSUD. Raising funds to promote research directed towards these goals is essential. Our participation in the 2018 Million Dollar Bike Ride sponsored by the University of Pennsylvania's Orphan Disease Center (ODC) was a major step in this direction, and we are thrilled that the MSUD Family Support Group has been invited to participate in the 2019 ride! This year's event is scheduled for June 8th, in Philadelphia, PA. Greater participation by our members will increase our reach, so spread the word

far and wide and share this link: 2019 MSUD Family Support Group And The Million Dollar Bike Ride .

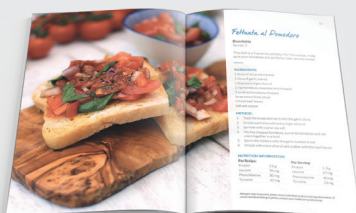
What We Accomplished In Last Year's MDBR

We had a very successful campaign last year. With our proceeds, we were able to fund a research project now underway at the Orphan Disease Center at the University of Pennsylvania. Under the leadership of Dr. Jenny Greig, Director for Liver Metabolic Diseases, the goal of this project is to develop a muscle directed gene therapy approach for MSUD

'Bike Ride' Continued on page 4

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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 has been busy since the 2018 summer symposium. We held quarterly conference calls in Sept 2018 and Jan 2019 and discussed numerous projects

including this year's participation in the Million Dollar Bike Ride and partnering with the Orphan Disease Center at the University of Pennsylvania (UPenn) to organize a Think Tank meeting with researchers and clinicians to identify how we should direct our research efforts. We are grateful for the support of the UPenn Orphan Disease Center. Read more about both of these projects in this edition of the MSUD newsletter.

Our biannual symposium is always something to look forward to, so start planning now! The next MSUD Symposium will be held in Columbus, Ohio on June 25-27, 2020. Once again, there will be inspiring topics and speakers and opportunity for fellowship.

We continue to fulfill our commitment to increase awareness of MSUD by participating in national conferences as advocates, exhibitors, and speakers.

We have a new board member! The board voted to invite our Advocacy Chair Jordann Coleman from CA and mother of Carter with Classic MSUD to join the board. We are thrilled that she accepted this invitation and know that she will be a great addition. With the addition of Jordann, the board has a diverse group of members with unique strengths and areas of expertise. I am fortunate to work with such dedicated individuals.

Please feel free to contact me anytime at 740-972-5619 or sandybulcher@gmail.com with your thoughts and ideas. And we can always use your help with our fundraising and advocacy efforts! ■

EDITOR'S NOTE



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

Welcome to the Spring 2019 issue of our newsletter!

Research, fundraising, and advocacy continue

to be areas of focus for us. I was pleased to be able to attend Rare Disease Advocacy Week in Washington, DC, where I made sure legislators are aware of MSUD and ways in which they can help improve the lives of those with this disease. See our Legislative Chair and new board member Jordann Coleman's article for more on how you can help our advocacy efforts.

We are getting into high gear for our participation in this year's Million Dollar Bike Ride. Board member Ed Fischler explains how you can participate, whether you're a cyclist or not.

Have you wondered how protein requirements are determined for those on an MSUD diet? Our nutrition section features an article by our advisor Dr. Sandy Van Calcar explaining how the need for adequate protein for growth is balanced with the amino acid restrictions we must adhere to.

As usual, we enjoy hearing from our families. We are delighted that Ethan Jackson's parents and Jaylani Gendron's mom have shared their stories with us.

Finally, a special thank you to Nutricia, Cambrooke Foods, and VitaFlo for their continued support.

Please feel free to send suggestions for future articles as well as your own family stories to me at any time. My email address is: karen.dolins@yahoo.com.

I hope you enjoy this issue! ■



'Bike Ride' Continued from front cover

and compare its effectiveness with liver-directed approaches. This project will be executed during 2019 and conclude late this year or early in 2020.

Why Are We Doing This Again?

Each research project is but a single step in the journey of finding improved treatments and a cure for MSUD. Our participation in the ODC's 2019 Million Dollar Bike Ride will allow us to continue to explore improved treatments and search for a cure.

How You Can Help

Volunteers are needed for the 2019 MDBR. As successful as we were in 2018, we accomplished that success with only seven families volunteering as riders and campaigners. Imagine what we could accomplish if everyone helped out! There are four ways you can volunteer and help us raise funds or donate.

To volunteer:

- 1. Join our team of riders in Philadelphia** – Last year, three families rode in the Philadelphia event on behalf of our team. We need more families to ride with us in Philadelphia on June 8th. The cost to register is just \$25 (note, you must raise at least \$250 before the event). Fundraising cyclists will receive an official 2019 Million Dollar Bike Ride cycling jersey and T-shirt.
- 2. Join our team as a "virtual rider"** – Last year, we had two families ride in their hometowns as virtual riders. We can do better! Gather a group of

friends or ride on your own in support of MSUD. You can ask your team members to contribute to the general MSUD fundraising page (see: <http://givingpages.upenn.edu/FamilySupportGroup>) or you can create your own individual page for contributions. There is no cost for doing either.

- 3. Join our team as a fundraiser** - Don't own a bike or nervous about riding 13+ miles? It's ok! You can still support our group by helping us fundraise. Fundraisers can create their own individual pages to help us reach our goal. You can customize your page with stories about you/your loved one's life with MSUD, pictures, and videos. There is no cost to being a fundraising-only participant.

Or, you can make a donation

- 4.** Not interested or don't have the time to run a fundraiser? You can still support us! You can either donate via the MDBR MSUD fundraising page or send a check to the MSUD Treasurer, with your contribution:

Dave Bulcher, MSUD Treasurer
9517 Big Bear Avenue
Powell, Ohio 43065

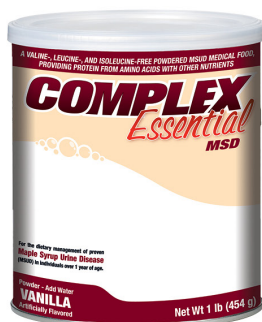
Questions?

If you wish to volunteer or have any questions, please contact Ed Fischler, VP of the MSUD Family Support Group Board of Directors at ebfischl@gmail.com. ■



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INTRODUCING OUR NEWEST MSUD FAMILY SUPPORT GROUP BOARD MEMBER - JORDANN COLEMAN

Earlier this year, The MSUD Family Support Group Board of Directors extended an invitation for me to join the Board and I happily accepted! During the day, I'm a risk advisor for a large regional insurance brokerage. By night, I'm a busy mom! My husband, Andre, and I live near San Francisco, California and have three children: two daughters ages 8 and 9 months and a son, Carter, age 6. Carter was diagnosed with Classic MSUD at 4 days of age through Newborn Screening. Today, Carter is in kindergarten, has lots of friends, and plays several sports.

I started working with the MSUD Family Support Group about 5 years ago when I volunteered to lead our advocacy efforts. I had gotten a little taste of advocacy with Newborn Screening in California and wanted to spend more time advocating on behalf of MSUD. For a little over a year, my advocacy focus has been on the Medical Nutrition Equity Act. In May, I will travel to Washington DC to advocate for its support and passage.



We would love to have our members set-up meetings with their Congressional Members when they are home for their summer recess in August to advocate for this important legislation. Rare Disease Legislative Advocates (RDLA) helps to prepare advocates for these meetings, providing legislative resource materials and hosting pre-meeting training webinars. If you're interested in participating, register between May 8th and July 3rd at www.RareAcrossAmerica.org. No prior advocacy experience is necessary. Contact me at coleman.jordann@gmail.com for more information. ■

NBS CONNECT REGISTRY CLOSING ANNOUNCEMENT



by Aileen Kenneson, PhD, MS
Associate Scientist
Emory University, Dept. of Human Genetics

We are sad to report the closure of the NBS Connect registry despite generous support from groups such as the MSUD Family Support Group, the growing costs of maintaining the registry have made it unsustainable. As a result, the NBS Connect registry website was closed on February 28, 2019.

The NBS Connect registry ran for seven years. Over 600 people were registered with NBS Connect, including about 100 people with MSUD and their families. Of these, 58 people completed the MSUD survey. The information in this survey was used to describe the natural history of MSUD, and a research paper on this topic was published in 2018.

The data that users entered into NBS Connect, including contact information, will be maintained on a secure server at Emory University. Only NBS Connect staff will have access to these data. In the event that another MSUD patient registry is started, your data will NOT automatically be shared with that registry. Rather, an email will be sent to you asking if you would like to have your information shared with the new registry. Your information will only be

shared with a new registry if you provide permission.

The aim of the MSUD survey was to provide valuable data to researchers to improve the lives of those with MSUD. Researchers will still be able to work with us to access the MSUD survey data if they are interested in it. They would need to complete an approval process, and sign a data use agreement with Emory University that states their

responsibilities to keep the data secure and private. Only de-identified data will be provided. That is, they would not have access to any data that could identify you, such as names, birthdates or zip codes.

From time to time, NBS Connect notified users of updates and other valuable information about NBS Connect and related clinical and research information. Your email addresses will be maintained so that additional information about NBS Connect and related information can be shared with you, if needed.

One part of NBS Connect that was of interest to users was the Kitchen Connection. This provided low-protein recipes complete with nutrition information (such as leucine content) as analyzed by registered dietitians. These recipes will be transferred to the Medical Nutrition Therapy for Prevention (MNT4P) Program website at MNT4P.org for inherited metabolic disorders, and we will continue to add to the recipe collection. The MNT4P program provides services for individuals with metabolic disorders and their families in the state of Georgia, but the recipes will be available for everyone.

We thank everyone who participated in the NBS Connect registry. ■

ADVOCACY

ADVOCACY UPDATE - MEDICAL NUTRITION EQUITY ACT



By: Jordann Coleman

The Medical Nutrition Equity Act ensures public and private insurance coverage for medically necessary foods when prescribed by a physician. The bill was introduced to the 115th Congress, however it was never brought to the floor for a vote. It will be re-introduced to the 116th Congress later this spring.

Lead Co-Sponsors for the House version of the Medical Nutrition Equity Act have been identified - Rep. Jim McGovern (D-MA) and Rep. Jamie Herrera Butler (R-WA). In the Senate, Sen. Casey (D-PA) has committed to being a lead co-sponsor, but a republican lead co-sponsor is needed.

In addition to the leads, other co-Sponsors in the US Congress and Senate are needed for the Medical Nutrition Equity Act. After the 2018 election, there are 24 co-sponsors on the House version

of the bill (7 Republican & 17 Democrat) and 4 co-sponsors for the Senate version of the bill (1 Republican & 3 Democrat).

We are asking our US-based members to contact their members of Congress (especially if they are Republican) and ask them to co-sponsor this bill. You can go to equity4.us/takeaction to send an auto-generated email to your members of Congress. You can also visit medicalnutritionequityfor.us to share why having coverage for medical nutrition is important to you and your family.

The Patients & Providers of Medical Nutrition Equity Coalition will be having a Hill Day in May

to generate more support for this bill. I will be attending as a representative for the MSUD Family Support Group and joining other advocates on the Hill. We would love to have our members set-up meetings with their Congressional Members when they are home for their summer recess to advocate for this important legislation. Contact me and I can help to set-up these meetings.

We need as many voices as possible to help us pass this bill. 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! ■

NEWBORN SCREENING SAVES LIVES REAUTHORIZATION ACT OF 2019

The original Newborn Screening Saves Lives Act was passed by Congress in 2008. This legislation established national newborn screening guidelines, including a Recommended Uniform Screening Panel identifying conditions to include in screening, and helped facilitate screening in every state.

This act must be reauthorized every 5 years. It was reauthorized in 2014 and must be reauthorized by September 30, 2019.

Please contact your legislators and encourage them to support reauthorization. Tell them your story and why Newborn Screening is vital to you. ■

RARE DISEASE WEEK IN WASHINGTON, DC:

ADVOCATING FOR MSUD*By Karen Dolins*

between states, and making recommendations for conditions to include in screening. This program must be reauthorized every 5 years. The current reauthorization bill is sponsored in Congress by Representatives Raybal-Allard and Simpson. The bill has not yet been introduced in the Senate. The proposed bill will increase authorization levels and commission a report by the National Academy of Medicine on modernization of the program. The deadline for reauthorization is September 30, 2019.

February 24-28 is Rare Disease Week, and I joined almost 500 other advocates in Washington, DC for conferences and meetings with legislators on Capitol Hill. This annual event is organized by Everylife Foundation's Rare Disease Legislative Advocates (RDLA) with the goal of giving patients a voice. The first day of meetings was designed to give us an understanding of the current climate in Washington and how best to achieve our goals.

A major focus of the current Congress is to lower the cost of prescription drugs, making them more affordable. This goal is shared by the Trump administration. Speakers emphasized that lowering costs must be accomplished by addressing excessive profits by the pharmaceutical industry, and not in a way that will discourage innovation for new drugs.

Three legislative asks were identified for 2019:

1. Newborn Screening (NBS) Reauthorization - we are fortunate that all states now screen for MSUD. While NBS is conducted by individual states, the federal government plays a role by providing technical assistance, facilitating collaboration

2. FDA and NIH appropriations - Increased funding for both the NIH and FDA is needed to meet the needs of the rare disease community. Due to increased costs, funding must increase annually just to maintain the status quo. The President's current draft of the Fiscal Year 2020 budget would decrease funding to each of these agencies by 15%, resulting in significantly fewer funds available for rare disease research.
3. Rare Disease Caucus – Legislators meet quarterly for updates on issues facing the rare disease community. This keeps our needs on their radar screen.

This was the second time I attended Rare Disease Week on Capitol Hill. After my previous visit, I followed up with my Congressman and was thrilled when he joined the Congressional Rare Disease Caucus!

Rare diseases enjoys bipartisan support and is considered a nonpartisan issue. There are over 7,000 rare diseases affecting an estimated 30 million Americans. By joining together in our advocacy efforts, our voice can be heard. ■

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FOOD AND NUTRITION

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the countryside of Tuscany. At that time, the process of making and drying the pasta was carried out in the courtyard under the sun. This all nearly ended during the Second World War, when the company was bombed and partially destroyed. After the bombing, the Mennucci family was quick to rebuild the factory. During

the rebuilding process, all pasta machines that were not destroyed were moved into local homes near Tuscany to maintain the production of pasta.

The Mennucci family are very passionate about using the highest quality of ingredients which are mainly sourced from Italy. There are only four ingredients in low protein pasta which are mainly starches; corn, potato, rice and coloring (annatto). Lorenzo



'Loprofin® Pasta' Continued on page 11

UNDERSTANDING PROTEIN NEEDS FOR MSUD



**By Sandy Van Calcar,
PhD, RD**

Protein is an essential nutrient, meaning it is needed for life. For someone on an MSUD diet, this protein comes from two sources:

1. Medical formula, which provides “protein equivalents” from all the amino acids which make up protein except leucine, valine and isoleucine.
2. Limited amounts of “intact” protein from foods (or regular infant formula for infants) to meet an individual’s leucine needs.

“Total” protein is a combination of protein equivalents from formula and intact protein from food. For many with MSUD, the medical formula provides a majority of their total protein needs since the need to restrict leucine allows for a limited amount of intact protein.

Recommendations for intake of protein for individuals of different ages are published in the “Dietary Reference Intakes” (abbreviated DRI), which are established by the Institute of Medicine in the U.S. These guidelines are intended to cover 97% of the population who do not have any health issues. Since MSUD requires such a specialized diet, metabolic dietitians do not rely solely on the DRI when establishing protein needs for their patients.

Why is this? Amino acids are metabolized in the gastrointestinal (GI) tract differently than intact protein from foods. This influences how much total protein is prescribed for those with MSUD. Intact protein from food must first be broken down (digested) into amino acids before it can be absorbed from the small intestine into the blood stream. This process is slower than absorption of the amino acids

in formula. Since amino acids in formulas are already “broken down”, they are more rapidly absorbed into the blood stream and are taken up by the body’s cells sooner than amino acids produced from intact protein in food. Studies have shown that retention and utilization of amino acids for body protein synthesis is better when a protein source is an intact protein compared with an amino acid source.

Given this difference in how protein and amino acids are utilized, it is common practice to prescribe total protein that is a minimum of 20 to 40% higher than the DRI for someone with MSUD. This helps to ensure that an individual will have a sufficient supply of protein in his/her diet for normal growth and body protein maintenance.

Below is an example of how protein is prescribed and distributed in an MSUD diet:

A 3-year-old girl weighs 30 pounds (14 kilograms or kg)

DRI for total protein for a 3 year old = 1.1 grams of protein per kg of body weight

Her requirements are then calculated as follows:

$14 \text{ kg} \times 1.1 \text{ grams protein} = 15 \text{ g of protein per day}$

We want a total protein that is 40% higher than the DRI so we multiply $15 \times 1.4 = 21 \text{ gram protein per day}$

Amount of leucine in this girl’s diet = 400 mg, as determined by monitoring blood levels. This is equivalent to about 7 g of intact protein from food. (There are about 60 mg of leucine in 1 gram of protein).

As her total protein prescription is 21 grams and she will get 7 grams from food, she needs 14 grams of protein equivalents from a medical formula.

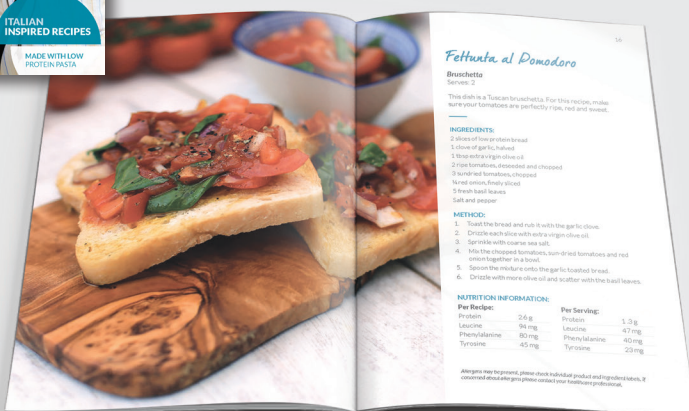
Calorie intake must also be adequate so that the amino acids from formula and food will be available for protein synthesis rather than broken down to supply energy. Most formulas for MSUD include both fat and carbohydrate sources to provide these needed calories ■

'Loprofin® Pasta' Continued on page 9

believes that when there are so few ingredients in a product it is important that you use the highest quality of ingredients.



View recipes made with Loprofin Low Protein Pasta at MedicalFood.com/Recipes



The main difference in the production of low protein pasta and ordinary pasta is that gluten is the protein that binds the dough in ordinary pasta. For low protein pasta, steam is used instead to bind the starches together to make a dough. Low Protein Pasta is made using spring water that comes from the Tuscan mountains. ■



See for yourself where our Loprofin Pastas are made in this video of the Mennucci family! Visit YouTube.com/Lowproliving



INSTANT POT PEACH JAM

(Recipe by Dana Angelo White, RDN)

MAKES 3 CUPS

- 5 cups fresh peaches (peeled, chopped, diced)
- 2/3 cup granulated sugar
- 1/3 cup wildflower honey
- 1 tablespoon lemon juice

Stir peaches, sugar and honey in the pot. Cover and lock the lid. Pressure Cook (high) for 2 minutes. Allow for natural release. Remove lid, stir in lemon juice. Use a potato masher or immersion blender to break down peaches. Simmer in saute mode 10 to 12 minutes until thickened. Cool before covering and refrigerating. Expires in 3 weeks.

NUTRITION INFO (1 TABLESPOON):

Calories: 48; Protein 0 g; Total Fat: 0g; Carbohydrate: 8g

<http://danawhitennutrition.com/2018/09/instant-pot-peach-jam/> ■

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MANGO FRUIT LEATHER

(Recipe by Dana Angelo White, RDN)

YIELD: 12 STRIPS

- 2 cups chopped fresh mango
- Juice of 1/2 a lime
- 2 tablespoons sugar
- 2 tablespoons agave nectar

Preheat oven to 170°F. Line a baking sheet with parchment paper and set aside. Place mango, lime, sugar, and agave in a medium saucepan. Bring to a boil and cook 7 to 8 minutes. Puree using an immersion blender or food processor. Cook over medium-high heat for an additional 5 to 10 minutes or until syrupy (e.g. thick enough to coat the back of a spoon). Pour mixture into an 8×12 inch rectangle on baking sheet. Bake for 3 1/2 hours. Turn off the oven and allow to sit overnight. Cut into strips with a pizza cutter; roll-up in a clean piece of parchment paper. Store for up to 3 weeks in an airtight container.

NUTRITION INFORMATION PER 1 STRIP

Calories: 36, Total Fat: 0 grams, Saturated Fat: 0 grams, Carbohydrate: 10 grams, Sugars: 6 grams, Protein: 0 grams, Sodium: 1 milligram, Fiber: 1 gram

<http://danawhitenutrition.com/2017/03/healthy-snack-mango-fruit-leather/> ■



RESEARCH

NAVIGATING THE PATH TOWARDS IMPROVED TREATMENT AND CURE FOR MSUD

By Karen Dolins

Those of you who read our newsletter and visit our website know that research is a major focus of our organization. We have provided financial support for various research studies throughout the years. Typically, researchers have approached us with requests for projects which they feel will help identify improved treatments for MSUD. If the board and our medical advisors agree, we provide the support.

We realize that we have been reactive rather than proactive, responding to requests rather than requesting proposals for research projects that we have identified as important. We hope to remedy this with a Think Tank – a meeting with key clinicians and researchers who will review current treatment strategies, identify potential novel approaches, and consider research needed to close the gap between



Photo by Alexander Andrews on Unsplash

what we now know and what we need to know to advance care.

We have been very fortunate in that the University of Pennsylvania's Orphan Disease Center has offered to partner with us in this endeavor. They will be hosting a meeting of key researchers and clinicians on June 20, 2019, which MSUD Family Support Group President Sandy Bulcher and I will attend.

We look forward to this opportunity to learn from the attendees and advance research in a meaningful way. ■

FROM THE MEDICAL JOURNALS

Summarized by Karen Dolins

MSUD often results in neurological damage. The breakdown products of leucine, isoleucine, and valine are implicated in this damage. It has been suggested that oxidative stress plays a role, and that supplementation with L-Carnitine help protect against this damage.

Carnitine is a substance with antioxidant and anti-inflammatory properties which is involved in the metabolism of fats. It is obtained from the diet and also made by the body. Some individuals who rely on a synthetic formula for their nutrition may develop a carnitine deficiency.

In one study, 7 individuals with MSUD were

supplemented with 50 mg L-Carnitine per kilogram body weight daily for 2 months. Urinary levels of 8-OHDG, a marker of DNA damage, were measured before and after supplementation.

The researchers found that the amount of leucine in the urine was correlated with levels of 8-OHDG, and that individuals who received L-Carnitine had lower levels of 8-OHDG after 2 months than at baseline. They theorize that supplementation may help reduce DNA damage and ultimately play a role in reducing neurological damage in individuals with MSUD.

Hauschild TC et al. DNA Damage Induced by Alloisoleucine and Other Metabolites in MSUD and Protective Effect of L Carnitine. *Toxicology in Vitro* 57 (2019) 194–202 ■

UNDERSTANDING GENE THERAPY



Image: Pixabay/geralt

Are you interested in learning more about gene therapy? The American Society of Gene & Cell Therapy (ASGCT) can help!

Visit their website at <https://www.asgct.org/education> for their easy-to-understand Gene Therapy 101.

ASGCT's members consist of scientists, researchers, clinicians and many other stakeholders involved in the

field of gene and cell therapy. Their patient education initiative began in 2018 to provide reliable, useful, patient-friendly resources on a rapidly advancing science. To create the content they collaborated with many patient advocacy organizations, foundations and industry representatives, along with the expertise of their own membership. ASGCT plans to continue these education efforts and focus on seven new disease areas this year. ■

UPCOMING EVENTS

SAVE THE DATE

MSUD Symposium 2020 will be held on June 25th-27th at the Embassy Suites Columbus Airport, Columbus, Ohio. Hope to see you there! ■



FAMILY NEWS

JAYLANI GENDRON

AGE 4 ½ YEARS, CLASSIC MSUD

By Jayna Gendron, mom



the doctor if my daughter was going to die was the most heartbreaking thing I had ever experienced. I remember being angry with God asking why did this have to happen to my child. Some days I still find myself angry with him that my perfect little angel has to go through so much and I cannot take her place so she doesn't have to endure the effects that MSUD has on her life.

Jaylani was born in October, 2014, a month early due to preeclampsia, and weighing just shy of 4 pounds. Due to her small size, she was placed in the NICU.

Jaylani was feeding normally until Day 5 of her life when I noticed she was no longer finishing her feeds. She was very sleepy which was normal for a newborn so we did not feel there was cause for concern. Later that day we received the news that her newborn screen had come back. Jaylani had MSUD and a metabolic doctor would be coming to talk to me. I immediately googled it to see what this meant for my daughter. The results were scary and very confusing, especially when I read that my child could die. After speaking with the metabolic doctor and getting an idea of what life would be like for Jaylani, I had to get a grip on the damage which needed to be reversed as her leucine was now 4900, well above the normal range of 100-300. Watching my little baby lay there basically comatose and with a breathing tube was devastating. Asking

Jaylani has had some hospitalizations where she has needed a NG tube to help her drink the amount of formula she needed. Through it all she is always smiling and a happy child. Looking back on those first few weeks when I didn't know if we would make it to today, where I have a beautiful, happy and loving 4 year old whose smile lights up my life, I can't imagine my days without her. She is truly a miracle. Today Jaylani attends preschool. She enjoys riding the bus as it makes her feel like a big girl, loves going to the park, and her favorite thing is to be on the swings no matter the weather.

We have a meeting scheduled with a transplant team to gather more information and see if this is an option that would be best for her.

Jaylani can be a little shy but she is gentle and loving and her smile brightens anyone who comes into contact with her. I am grateful every day that she chose me to be her mother. Thank you for this opportunity to share my daughter's journey with you. ■

JUST A BOY

WHO HAPPENS TO HAVE MSUD



***By John and Patti Jackson
Parents of Ethan, Classic
MSUD Age 14***

So much time has passed since we first took on the mission to ensure our son Ethan always had a place at the dinner table. Roughly a lifetime ago, it seemed our life was

consumed with counting, weighing, monitoring and logging every gram of food he ate. We spent time worrying, praying, and seeking glimpses of light while trying to avoid shadows of darkness when possible. We spent hours - even days - checking levels, desperate for normalcy and, of course, our prayers continued.

Ethan was on the precipice of being tagged an MSUD patient named Ethan rather than a boy named Ethan who happened to have MSUD. We quickly learned that we had to treat him as our son, not as our patient, if he was going to have a chance to live his life to the fullest. Our realization was the moment when Ethan's potential became boundless.

Ethan's life transformed from a singular prism of triage to limitless possibilities and endless achievable dreams. This revolution set a trajectory for Ethan, which has been steadfast ever since, and opened the doors to the best possible opportunities of his choosing. No longer would he be consumed with MSUD in a way that hindered his state of mind. He smiles when he is happy, he cries when he is sad, and he acts out when adolescence dictates he should. He is everything a 14-year old should be.

He has discovered talents which he has turned into activities he enjoys to do, like baseball. He's found a passion for film editing, which in turn has sparked a

drive to one day make this his career.

He has plans.

Let that sink in as it has for us.

He has plans for a future of his making. This again supports this thought of MSUD being what he has, not who he is. Maple Syrup Urine Disease is part of who he is, and the person he is striving to be every day. He expresses every aspect of who he is. It is amazing to see how, for Ethan, MSUD is a way of life worth embracing. From researching MSUD for school projects to learning about the science behind it for his own thirst for knowledge, science, technology and mathematics have become the foundations of Ethan's educational journey.

The 3rd grade introduced Ethan to the world of STEM (Science, Technology, Engineering, and Mathematics). In the beginning we did not think this was even in the realm of possibilities for him but STEM became as much of who Ethan is as MSUD. Education (even though, if asked, he would say he's not a fan) provided Ethan with an outlet for all his wonderment, need for inclusion, and faith in who he is being enough. He built friendships with people who accepted each and every part of who he is. Questions about MSUD became a part of daily conversations, and he took pride in his replies. Ethan was in control of who he let in, and how he wanted his story to be told.

Today, Ethan is in the middle of his freshman year of high school. His landscape has changed from his five years at STEM, but he carries with him a solid roadmap to help guide him through the most socially diverse environment thus far. He has kept in touch with old friends while creating new relationships. He excels in his academics while cultivating his pre-adult social skills.

Ethan has a gift. He didn't ask for it, he certainly did not want it, and at times had wished it would just go away. Today we still count, weigh, and monitor every gram of food he eats. We spend time worrying, praying, and seeking glimpses of light while trying to avoid shadows of darkness when possible. But here is the biggest difference between today and yesterday: Today, Ethan is just a boy who happens to have MSUD (Period). ■