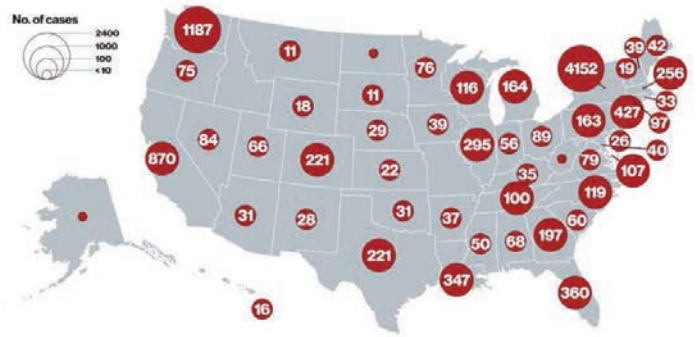




MSUD NEWSLETTER

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Current as of March 19, 2020; ABC News

OPEN LETTER FROM THE CLINIC FOR SPECIAL CHILDREN

Dear Friends,

Ensuring your safety and well-being is at the heart of our mission at the Clinic for Special Children (CSC). In that spirit, we are taking steps to protect you and your community from **COVID-19**. I am writing to provide critical information about the virus, what it means for you, and how we can work together to protect our loved ones.

What is the COVID-19 Pandemic?

Coronaviruses commonly cause respiratory illness similar to influenza (“the flu”). In November 2019, a particularly dangerous strain of coronavirus called COVID-19 first appeared in China, and over just a few

months has spread rapidly across the globe to infect 250,000 people and cause 10,000 deaths (World Map, **red**). The virus reached the United States in January 2020, and as of March 20th has infected more than 15,000 people in all 50 states. There are multiple centers of contagion along the Eastern Pennsylvania border, and now a few confirmed cases in Lancaster County (U.S. Map, **red circles**).

COVID-19 spreads quickly for three reasons:

(1) It readily passes between people by either direct contact or shared objects (e.g. toys, hand towels,

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

MSUD FAMILY SUPPORT GROUP NEWS

FROM THE
PRESIDENTS' DESK

By Sandy Bulcher

By now I'm sure you've heard that the MSUD Symposium scheduled for this June has been cancelled. Our symposiums play a significant role in connecting families and increasing our knowledge of MSUD,

and we are saddened that we will not have these opportunities. The symposium committee had been planning for the event for months and the agenda was packed full of great speakers and topics. While I'm very disappointed that the symposium had to be cancelled, it was not a difficult decision to make considering the vulnerability of our population and the uncertainty of the Covid-19 virus.

The MSUD FSG leadership will be discussing our options and evaluating how we can best provide information and foster networking during these uncertain times. We will update our membership when this has been determined.

The Covid-19 crisis certainly has changed our lives and caused increased anxiety over work challenges, finances, and childcare for many of us. Besides those typical stressors, our community has additional worries regarding the health and safety of those with MSUD and liver transplants. Please know that the MSUD leadership is committed to doing what we can to support the MSUD community and encourage you to continue supporting each other. We've always been a close knit group and can get through this together. Check out the resources in this newsletter for more information. Stay well! ■

EDITOR'S NOTE



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

Welcome to the Spring 2020 issue of our newsletter!

When I first began preparing this issue of our newsletter in January, Covid-19 was something that was happening far away and hadn't yet touched our lives. Now I write this from the confines of my home just north of New York City, where we have an abundance of cases. I, like all of you, am consumed by thoughts of how to keep my MSUD child safe.

This newsletter is our primary means of communication. This issue includes the family stories that we read with empathy, information on tools we can use to make the MSUD diet easier to follow, a summary of an important study conducted by the Clinic of Special Children in Pennsylvania, and advertising from our sponsors. We report on board activities in the areas of research, fundraising, and advocacy. In this issue we also include resources about avoiding Covid-19 from scientists whose job it is to keep us all safe.

We are planning to continue our communication with you, our community, through regular eblasts and posts on our official Facebook page. As always, if there is something you would like us to include in our next issue, please contact me at karen.dolins@yahoo.com.

Yours in health. ■

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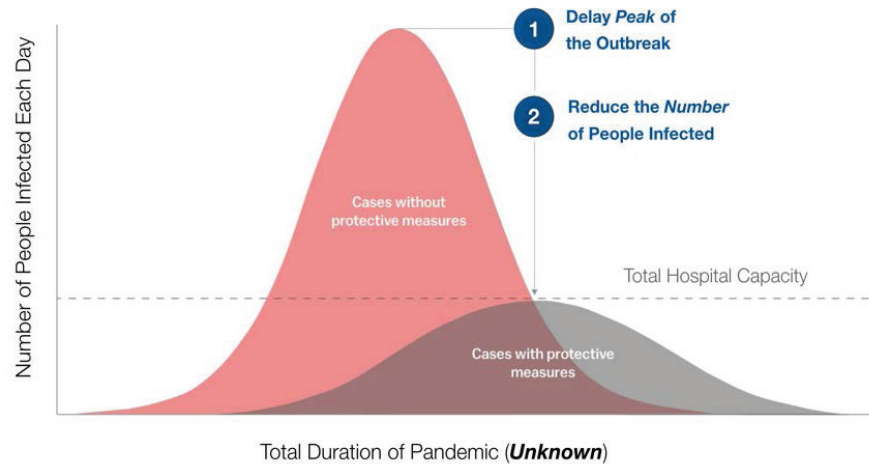
'Clinic' Continued from front cover

dinner plates), and is so contagious that it can be passed from one person to another through normal breathing; (2) It is a novel virus, never before seen on Earth, which means that no one is immune to COVID-19 and nearly everyone who comes in contact with it will get sick; and (3) Once infected, a person typically has no symptoms for 7–14 days, during which he or she unknowingly infects others, who in turn pass it on to their close contacts, and so on.

COVID-19 is not “the flu.” Although COVID-19 has been compared to influenza virus, there are three important differences between them: (1) Unlike the flu, which typically infects people during a few winter months, the COVID-19 pandemic could last 18 months or longer; (2) We have methods to prevent (vaccines) and treat (medications) the flu. In contrast, there are currently no effective treatments for COVID-19; and (3) COVID-19 is much more deadly than the flu. A person infected with COVID-19 is 15–times more likely to die than one infected with the flu, especially if elderly or medically fragile.

Understanding the pace of the pandemic is important. As of this writing, the number of people in the U.S. who become infected doubles every three days. This means that if 15,000 people in the U.S. have the virus today, 30,000 will have it in three days, 60,000 in six days, and so on. Without protective measures, the entire U.S. population (330 million people) could be infected in less than six weeks. In just nine weeks, the virus could spread to every person on Earth.

What does this mean for the medical system? There are one million hospital beds in the U.S. and about 70% of them are occupied at any given time. One in six people who contract COVID-19 need to be hospitalized. This means that at the current rate of spread, every hospital bed in the U.S. will be filled by April 10th, 2020, just three weeks from now. Once this happens, doctors will not only run out of critical supplies and resources, but will be forced to make heart-breaking decisions about who does and does not receive medical care. People who do not have COVID-19, but suffer from other life-threatening conditions, may be excluded from hospitals operating at maximum capacity.



How Can We Control COVID-19?

Although there is no treatment for COVID-19, protective measures can limit its impact on communities worldwide (Figure modified from CDC, below). In the absence of protective measures, the number of people infected (red shaded area) quickly overwhelms total hospital capacity (gray dashed line). Implementing protective measures early in the course of the pandemic (gray shaded area) has two effects: (1) The peak of the outbreak is delayed, giving medical systems more time to respond; and (2) Fewer people become infected.

What Can You Do to Help?

- Wash your hands often with soap and water for at least 20 seconds, especially after visiting with people outside the home, shaking hands, using the bathroom, or before preparing food.
- Avoid touching your eyes, nose, or mouth with unwashed hands.
- Avoid shaking hands and sharing hand towels.
- Unless necessary, avoid close contact with people who are sick.
- If you or a family member is sick, stay home unless you need a doctor's attention.
- Cover all coughs and sneezes. If you use a tissue, throw it away. If you use your hand, wash it.

- Frequently disinfect common objects and surfaces in your home.
- Avoid large, non-essential gatherings, even if everyone appears healthy.
- Respect the government recommendation to cancel all public meetings and limit any gatherings to fewer than 10 people. In Plain communities, this might include cancelling congregational worship, limiting marriage ceremonies to a few family members (could do meals and receptions at a later date) and keeping schools closed until the peak of the pandemic has passed and the government lifts restrictions. Countries that observe such recommendations have fewer infections and fewer deaths.

Protective measures save lives. This is our time to work together to protect our neighbors, our loved ones, and communities throughout the world. Until COVID-19 runs its course, I encourage you to stay calm, stay safe, stay clean, and stay home.

Yours Sincerely,

Kevin A. Strauss, M.D.
Medical Director, Clinic for Special Children

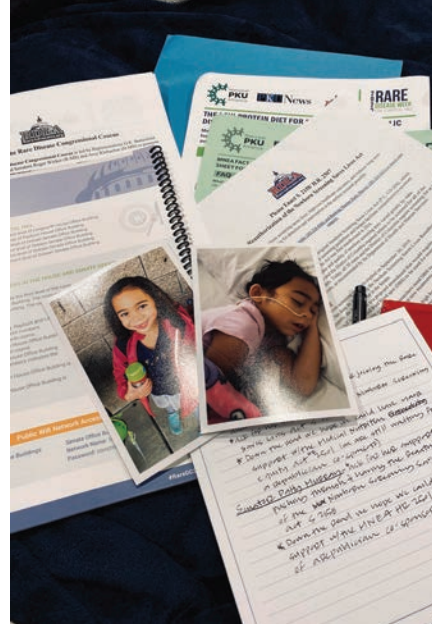
RARE DISEASE WEEK 2020

By Susan Mays



In February I had the opportunity and pleasure of traveling to Washington DC and attending Rare Disease Week. Rare Disease Week is an annual multi-day event hosted by the Everylife Foundation that is always held on, or close to, Rare Disease Day (last day of February). During Rare Disease Week patients and advocates from around the country have the opportunity to meet and network with others, learn more about becoming an effective advocate, hear about how other rare disease organizations are moving forward in connecting and supporting one another while pursuing a cure or better treatment options, and meet with their representatives and senators with specific asks (what we would like them to do) for their rare disease.

There is some variation from year to year as the event grows in content and attendance. Some of the highlights from my trip this year included a documentary screening called *Tin Soldiers*, a full day of seminars which helped me learn how to tell my story and articulate my asks, a chance to share and educate so many people about MSUD, and of course a chance to meet with the people within



government that are in a position to help make a difference in our lives.

This year I spent my time and energy advocating for the Newborn Screening Saves Lives Reauthorization Act, the Medical Nutrition Equity Act, and asking my representatives to join the Rare

Disease Congressional Caucus. Each of these bills has or could have a huge impact on our community. For me, Newborn Screening allowed us to get a timely diagnosis of MSUD in my newborn daughter, allowing her to begin treatment in a timely manner. The Medical Nutrition Equity Act would allow all families, in every U.S. state regardless of age/gender/socioeconomic situation, to have access to life saving medical foods and formula. Finally, the rare disease congressional caucus allows congresspeople and senators to be in touch with the needs of the rare disease community. This is a big step in showing their partnership and commitment to us.

I was so grateful for the chance to represent MSUD alongside so many other rare disease advocates and patients. Never in my wildest dreams did I ever think I'd be equipped to do this or have the chance to, but I'm so thankful I took that step and did it.

Now my challenge to you is to consider how can YOU take the first step in doing some advocacy work! How might you help raise awareness about MSUD? ■

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NUTRICIA

Maple Syrup Urine Disease

MSUD

Information for families after a
positive newborn screening



Adapted by the Dietitians Group BIMDG

BIMDG

British Inherited Metabolic Diseases Group



BASED ON THE ORIGINAL TEMPLE WRITTEN BY
BURGARD AND WENDEL

Reviewed & revised for North America by: A. Huber

This version of the TEMPLE tool, originally adapted by the Dietitians Group of the BIMDG for use within the UK and Ireland, has been further adapted by Nutricia North America for use within United States and Canada. This version no longer represents clinical or diagnostic practice in the UK or Ireland.

TEMPLE



Tools Enabling Metabolic Parents Learning

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Supported by NUTRICIA
as a service to metabolic medicine

For more educational tools, visit MedicalFood.com

Are you a new parent or relative learning about your baby's MSUD diagnosis? Nutricia North America's TEMPLE educational video series can help by explaining the disorder and its management. Pass it along! <https://www.youtube.com/watch?v=DkAVNQBK6ul>

*Editor's note: While this useful video mentions only Nutricia products, MSUD products are available through other companies.

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 - Red and Orange flavors
 - Suitable from 3 years of age

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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 originally introduced in 2018, during the 115th session of Congress, but never made it to the floor for a vote. The bill was reintroduced into the 116th Congressional session on May 2, 2019 and continues to gather more support. The House Bill (HR2501) currently has 60 co-sponsors (50 Democrats and 10 Republicans) and is led by Representatives Jim McGovern (D-MA) and Jamie Herrera Butler (R-WA). The Senate version of the bill is close to being reintroduced in the Senate as Senator Joni Ernst (R-IA) has agreed to be the lead sponsor along with Senator Bob Casey (D-PA).

Your action is needed! Due to the coronavirus pandemic, the Medical Nutrition Equity Act Hill Day scheduled for May 19, 2020 will now be a virtual grassroots campaign. On May 19th, join the MSUD

Family Support Group US-based membership by gathering friends & family to contact your Senators and Representatives to ask for their support of the Medical Nutrition Equity Act. Calls, emails, and tweets will be needed to obtain more co-sponsors for this important bill. Check with MSUD Family Support Group website and official Facebook page for more information as the date draws near.

If you are unable to make the virtual Hill Day on May 19th, you can ask your elected officials for support today. 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.

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! ■

RARE DISEASE DAY AT CODEXIS

By: Jordann Coleman

On Friday, February 28, 2020 I had the pleasure of serving on a panel in celebration of Rare Disease Day for Codexis, a protein engineering company based in Redwood City, CA. Codexis is currently in the process of developing a biotherapeutic treatment for PKU. The treatment Codexis has engineered for PKU is an active, therapeutic enzyme candidate which doesn't get broken down in the gastrointestinal tract. It is administered orally and compensates for the absence of the patient's missing natural enzyme by removing phenylalanine from the body. While this treatment is still in clinical trials, there is hope the same type of treatment may work for other inborn errors of metabolism (IEM) including MSUD.

The event kicked off with a presentation on IEMs from Dr. Gregory Enns, the head of Biochemical Genetics at Stanford (who is also my son Carter's doctor). Two other doctors from Stanford gave presentations on the science aspects of rare diseases. The second half of the event featured me, a family with a teenage daughter who has PKU, and a woman with Homocystinuria. The patient perspective was all about life with an IEM. The PKU family brought a variety of low protein foods, scales, and a food journal to show the reality of living on a low-protein diet.



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The woman with Homocystinuria described her life after being diagnosed at age 10 and relearning how to eat a low-protein diet after a decade on a normal diet. I shared with the audience the severity of MSUD and how quickly neurological damage can happen during a metabolic crisis. The Codexis employees in the audience asked insightful questions and many people came up to us afterward to thank us for sharing the reality of rare disease life. They were touched to see how the work they are doing in the biotherapeutic division can have a real impact on people's lives.

The event at Codexis was a great opportunity to show others how we live day in and day out with metabolic diseases. Vitaflo contributed to the event by setting up a tasting table so attendees could sample their products, including their metabolic formulas. I overheard several people say they were surprised how palatable the formulas were. I left the event very encouraged about the future of treatments for MSUD, and mindful of how important it is to share our stories. ■

THE MILLION DOLLAR BIKE RIDE

JUNE 13, 2020

**By Herb Foster, 2020 Team
Scott Leader**

My name is Herb Foster but my friends call me Butch. I serve on the Board of Directors for the MSUD Family Support Group. I have been asked to step in as Team Leader for this year's Million Dollar Bike Ride (MDBR) sponsored by the University of Pennsylvania's Orphan Disease Center (ODC).

First of all, I hope this newsletter finds all of our MSUD community safe and healthy during these very difficult times. The MSUD Board of Directors is focused on the current events pertaining to the Coronavirus and are here to help in any way we can.

Second, I would like to thank Ed Fischler for his leadership and hard work in getting our MSUD community involved with the MDBR and being our Team Leader for 2 years. Without his leadership and dedication, we would not be making the progress with research that we are. Thank you from the bottom of my heart, Ed, for all you do for the MSUD Family Support Group.

I along with my brother-in-law Steve Healy, Taryn Kessel and her husband, and Karen Dolins (another very dedicated board member) and her whole family have traveled to Philadelphia for the past two years to participate live and represent our organization. Many more riders across the country and in Canada rode locally in their communities for our MSUD team. It has been a great experience to come together with 900-1000 cyclists to ride for our respective Orphan diseases and raise money for our respective research projects.



This year the ODC has announced that the bike ride will be an all-virtual event.

Here they explain what this means for cyclists:

- Although we won't be seeing you in June for a 13, 34, or 72-mile ride, we ask and encourage you to ride on your own (or safely socially-distanced from your cycling buddy), between now and June 13th, and document your progress! We are looking into various cycling apps that will allow for progress tracking and sharing, allowing you to cycle at your leisure over the course of the next few weeks (or all at once!) Regardless of whether or not you choose to use the app that we suggest (or one of your own) we hope you will participate leading up to and including June 13th to show your commitment and spirit for the MDBR.
- Have a stationary bike at home? Cover your miles from the comfort of your home, and share your

status and progress with us along the way!

- Whether you're on your stationary bike or on the road, we will have special hashtags for sharing on social media, and ask that you post your personal or team cycling updates throughout the next several weeks up to and including June 13th. We will be sharing, "liking" and re-posting your updates on our social media channels and website, as well.
- As always, ALL registration dollars will be contributed to each team's grant fund, to help supplement your fundraising efforts. With that said, if you no longer wish to participate in the bike ride given the circumstances, please reply to me directly to process a refund. However, we encourage you to consider your registration fee as a (much needed) donation to your team's research grant.

The MSUD Board of Directors and I appeal to everyone in the MSUD community to make an effort to get involved in this year's bike ride and help us raise much needed funds for research to find a cure and improved treatments for MSUD. Your participation and donations are greatly appreciated. We are a small group but together we can make a difference.



My wife Diane and I have been involved in the MSUD Community for almost 49 years since our son Scott was born. Our MSUD team is named Team Scott in his honor. Like many other families, we are witnessing much progress and hope in our fight for a cure and better lives for our children and loved ones with MSUD.

I would like to thank the Orphan Disease Center at the University of Pennsylvania for all they do and for inviting our organization to participate again for the third year in the bike ride. They match up to \$30,000 of the money we raise and oversee the research projects we generate through our fundraising efforts.

Please join us virtually and make this year's event our most successful. ■

NOW ACCEPTING FACEBOOK DONATIONS!

If you have a Facebook page, you can help raise money for the MSUD Family Support Group! We are now one of the verified charities you can choose when creating a fundraiser through Facebook. Follow these simple directions to create your own Facebook fundraiser:

1. Click Fundraisers in the left menu of your News Feed
2. Click Raise Money
3. Select MSUD Family Support Group

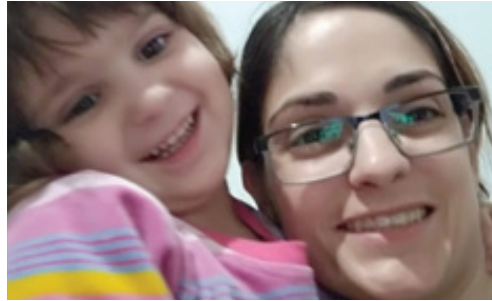
4. Select a charitable organization, choose a cover photo and fill in the fundraiser details
5. Click Create

Since our approval this past fall, we have raised over \$2,100 via Facebook! Fundraisers can be created at any time. Facebook users often create fundraisers for their birthdays or large charitable events like Giving Tuesday (the Tuesday after Thanksgiving). Next time you log into your Facebook account, try creating a fundraiser. Every dollar raised brings us closer to better treatments and a cure for MSUD. ■

ALÍA SÁEZ GIL

CLASSIC MSUD AGE 5 YEARS

By Aurora Gil Navarro, Mom



ALÍA SÁEZ GIL

CLÁSICO MSUD EDAD 5 AÑOS

Por Aurora Gil Navarro, mamá

This article has been translated from Spanish using Microsoft translate.

Hi, I'm Aurora. I have a 5-year-old daughter named Lya with MSUD. We are from Spain and this is our story:

Our story begins with a normal pregnancy with regular checkups and a normal delivery on March 27, 2015.

After 40 weeks of gestation and 8 hours of delivery, Alia was born. A healthy girl with big cheeks, she was perfect, except she didn't stop crying the whole time we were in the hospital. We were told it was normal because I was hungry. At the hospital, we gave her a bottle of milk to see if that way she would stop crying. She did, but not because she no longer starved, but because she started to get intoxicated.

When we were discharged we were told that in a few days we should do the heel test at our health center and we did. I have to say that in Spain depending on the Community in which you live, some diseases are tested for and others are not. Unfortunately, where we live Maple Syrup Urine Disease is not detected.

'Alía' Continued on page 13, column 1

Traducido del español con Google Translate

Hola, soy Aurora. Tengo una hija de 5 años llamada Alía con MSUD. Son de España y esta es nuestra historia:

Nuestra historia comienza con un embarazo normal con chequeos regulares y un parto normal el 27 de marzo de 2015.

Después de 40 semanas de gestación, Alía nació después de 8 horas de parto. Una niña sana con grandes mejillas, era perfecta, excepto que no dejó de llorar todo el tiempo que estuvimos en el hospital. Nos dijeron que era normal porque tenía hambre. En el hospital le dimos un biberón de leche para ver si de esa manera dejaba de llorar. Ella lo hizo, pero no porque ya no tuviera hambre, sino porque comenzó a intoxicarse.

Cuando nos dieron de alta nos dijeron que en unos días deberíamos hacer la prueba del talón en nuestro centro de salud y lo hicimos. Tengo que decir que en España dependiendo de la Comunidad en la que vivas, algunas enfermedades se contemplan y otras no. Desafortunadamente, donde vivimos la enfermedad de Jarabe de Arce no se detecta.

Después de unos días comenzamos a darnos cuenta de que Alía estaba muy dormida, apenas comía y respiraba extrañamente (pensamos que podría estar empezando a coger un resfriado).

En el sexto día de vida, la preocupación aumentó porque

'Alía' Continued from page 14

'Alía' Continued from page 12

After a few days we began to realize that Alía was very sleepy, barely eating and breathed strangely (we thought she might be starting to catch a cold). On the sixth day of life, the concern increased because Alía had not opened her eyes for many hours and barely ate, so we decided to take her to the health center. At first we were told that nothing happened to her, only that she slept a lot, but that we had to take her to the hospital since she did not eat and began to lose weight.

Upon arriving at the hospital, the doctor saw that Alía made strange arm movements (such as boxing) and became very rigid, suggesting a neurological problem. That afternoon she was admitted and the worst stage of our lives began. Alía was given test after test to rule out diseases because they did not know what was happening to her and she was getting worse hour after hour. At each consultation we were told that she was getting much worse and that several parts of her body were affected, so if she woke up there would be significant aftermath.

On the third day of admission and after countless tests came the best news. At last they knew what Alía had. It was a rare disease called Maple Syrup Urine Disease, and if she was looked after carefully there was a chance that the affected parts of her body would recover. And thanks to her excellent doctors, they transferred us to the Hospital La Fe of Valencia to be able to perform an emergency dialysis so that she could detoxify as soon as possible.

We had never heard of this type of metabolic disease, nor of the treatment, so we began an uninterrupted search for information, associations, food, etc. Here in Spain there are not many cases of MSUD so it has been very difficult for me to understand this disease.

We stayed in the hospital for a month because Alía was slow to wake up due to all the medication she was taking, and they had to perfect her diet before we could go home. Her blood tests at the time were done every week. Now that she is older she has blood tests every three months. Alía has great

doctors and dietitians, and has not had a serious decompensation since those early days.

Like every boy and girl with MSUD, her diet is based on fruit, vegetables, low-protein foods and a special formula that you have to take daily. In Spain the formula is subsidized by the government and thanks to that, so far we have never lacked it, since it is a very expensive product. The pharmacy takes care of ordering and supplying it.

At first, especially the first year, there was a lot of uncertainty since there were many revisions, a lot of tests to know if she had any continuing problems, and we did not know if we were doing everything right or wrong. But time creates a tranquility and makes you see how strong and hard these children are.

One of our concerns was the beginning of school (she started it when she was three years old) because we didn't know if she was going to be socially accepted and if she was going to be responsible enough not to eat forbidden things. We have been very surprised. She is very mature for her age, is able to say "NO" to all kinds of foods they offer her, gets along very well with all her peers, and academically is on the same level with them.

The problem with this society is that when you see that you can't eat many foods you think that "poor little girl" and do not realize that they are normal children, only they do not eat meat, fish, etc. It takes a great deal of empathy and inclusion.

Alía has an older brother (he doesn't have the disease) who has helped her a lot. Since she wants to do everything her brother does, this has pushed her neurologically.

Today, after almost 5 years, I have only words of thanks to the doctors who have helped us a lot, guided us and reassured us on numerous occasions. I am also SUPER proud of the person who Alía is becoming, of her responsibility, of everything she has taught us (you must never give up, you must always keep moving forward) because she is quite a warrior.

I don't know what the future holds, but as I once told you, "I took your hand so I'll never let go." ■

'Alía' Continued from page 12

Alía no había abierto los ojos durante muchas horas y apenas comía así que decidimos llevarla al centro de salud. Al principio nos dijeron que no le pasaba nada a la niña, sólo que dormía mucho, pero que debíamos llevarla al hospital ya que no comía y empezaba a perder peso.

Al llegar al hospital, el médico vio que Alía hacía movimientos raros con los brazos (como boxeo) y se quedaba muy rígida por lo que parecía un problema neurológico. Esa tarde fue ingresada y comenzó la peor etapa de nuestras vidas.

Alía pasó prueba tras prueba para descartar enfermedades porque no sabían lo que le estaba pasando y ella estaba empeorando hora tras hora. En cada consulta nos dijeron que estaba empeorando mucho y que varias partes de su cuerpo estaban afectadas, así que si se despertaba habría secuelas importantes.

En el tercer día de ingreso y después de innumerables pruebas llegó la mejor de las noticias, por fin sabían lo que tenía Alía. Era una enfermedad rara llamada enfermedad de la orina de jarabe de arce, y si se atendía rápidamente había posibilidades de que las partes afectadas de su cuerpo pudieran recuperarse. Y gracias a sus excelentes médicos, nos trasladaron al Hospital La Fe de Valencia para poder realizarle una diálisis de urgencia para que pudiera desintoxicarse lo antes posible.

Nunca habíamos oído hablar de este tipo de enfermedad metabólica, ni del tratamiento, por lo que comenzamos una búsqueda ininterrumpida de información, asociaciones, alimentos, etc. Aquí en España no hay muchos casos de MSUD por lo que ha sido muy difícil para mí entender esta enfermedad. Alía cuenta con grandes médicos y dietistas, y no ha tenido ninguna descompensación grave desde aquellos primeros días.

Nos quedamos en el hospital durante un mes porque Alía tardó en despertarse debido a todos los medicamentos que llevaba, y tuvieron que perfeccionar su dieta antes de que pudiéramos volver a casa. Sus análisis de sangre en ese momento se hacían todas las semanas. Ahora que es mayor se le realizan análisis de sangre cada tres meses.

Como todos los niños y niñas con MSUD, su dieta se basa en frutas, verduras, alimentos bajos en proteínas y una fórmula especial que tiene que tomar a diario. En España la fórmula está subvencionada por el gobierno y gracias a ello, hasta ahora nunca nos ha faltado, ya que es un producto muy caro. La farmacia se encarga de pedirlo y suministrarlo.

Al principio, especialmente el primer año, había mucha incertidumbre ya que había muchas revisiones, un montón de pruebas para saber si le había quedado alguna secuela, y no sabíamos si lo estábamos haciendo bien o mal. El tiempo crea una tranquilidad y te hace ver lo fuertes y duros que son estos niños.

Una de nuestras preocupaciones fue el comienzo de la escuela (ella comenzó cuando tenía tres años) porque no sabíamos si iba a ser aceptada socialmente y si iba a ser lo suficientemente responsable como para no comer cosas prohibidas. Nos hemos sorprendido mucho. Es muy madura para su edad, es capaz de decir "NO" a todo tipo de alimentos que le ofrecen, se lleva muy bien con todos sus compañeros, y académicamente está al mismo nivel que ellos.

El problema con esta sociedad es que cuando ven que no puede comer muchos de los alimentos nuestros piensan que "pobre niña" y no se dan cuenta de que son niños normales, sólo que no comen carne, pescado, etc. Hace falta un gran trabajo de empatía e inclusión.

Alía tiene un hermano mayor (no tiene la enfermedad) que le ha ayudado mucho. Como quiere hacer todo lo que hace su hermano, esto la ha ayudado neurológicamente.

Hoy, después de casi 5 años, sólo tengo palabras de agradecimiento a los médicos que nos han ayudado mucho, nos han guiado y tranquilizado en numerosas ocasiones. También estoy súper orgullosa de la persona en la que Alía se está convirtiendo, de su responsabilidad, de todo lo que nos ha enseñado (nunca debes rendirte, siempre debes seguir adelante) porque es toda una guerrera.

No sé qué nos depara el futuro, pero como una vez te dije pequeña guerrera "Cogí tu mano para no soltarla jamás". ■

ACHIEVING THE IMPOSSIBLE

JOEL FELSENSTEIN

CLASSIC MSUD, AGE 28 YEARS

With sheer determination, courage and strength I defied the odds and cheated death. I was born on January 2, 1992 in London, England with a very rare metabolic disease known as Maple Syrup Urine Disease (MSUD). The disease is so rare in fact that the doctors were almost 100% sure that I didn't have it, but after running blood tests the diagnosis was confirmed when I was 12 days old.

I went by police escort in an ambulance to Great Ormond Street Hospital in London while my parents followed in a car behind because they were not allowed in the ambulance.

I spent seven long months in Great Ormond Street Hospital in the ICU with IV drips, oxygen and various other tubes connected to me. The doctors said I'd never eat, talk, walk, or live anything like a normal life because of the extent of damage done to my brain caused by MSUD. By the general rule of science and medicine I should not be here now. The fact that I am is nothing short of a miracle. Between the ages of 4 & 18 I was an ostomate. I had a gastrostomy tube to deliver my medical formula.

Having this kind of condition doesn't bring friendships easily, and it's been difficult until now to share my story. I was bullied and lucky even to end up with one friend. I've had a hard time making friends and having relationships, but I have never



given up with life and have often fought to get what I wanted from life.

Even though the doctor's expectations of me were low, I started taking on every obstacle life threw my way and kept on smashing my way through them, achieving my goals and defying everyone's expectations of me! At 2 years old I had already decided that I would learn the drums and was banging everything. This was the start of my 26 year+ music career.

I have always had that determination from when I was born right up to today, and at age 28 I am still making my own path. I have met many awesome people along the way, from playing drums in the school band to performing with some amazing musicians both Jewish and secular. I currently live with my parents, not because that is my choice but because since losing my job back in October and becoming an entrepreneur I could not afford to move out on my own. When I do move out I am looking into moving away from the UK to California to pursue my American dream.

While music will always be a big part of my life, I will be taking on more challenges head on and pushing them out of my way so I can continue along the highway of life! I have started creating my own business with the single goal of helping people who think outside the box and have creative minds, from people with high functioning autism to budding entrepreneurs and others who are slightly disabled or at a disadvantage. I hope to change the business world forever!

What new and exciting challenges & adventures are there in store inside my world? If you'd like to join me on the Journey to find out, please follow my blog! You can email me at: joelfelsenstein@gmail.com ■



ISRAELLE THREET CLASSIC MSUD AGE 6 YEARS

By Lisa Alexander Threet, Mom



Israelle is my adopted daughter. She and her brother Isaiah first came to my husband Randy and me as foster children on our wedding anniversary.

Israelle was identified with high leucine levels through newborn screening in Tennessee 3 days

after her birth on February 19, 2014. Hospital officials tried to contact her birth family to advise them of the positive results, but they were unable to locate them. By the time Israelle was found, she was in a coma. Her parents had not sought care for her nor recognized the severity of her condition. She was immediately brought to the NICU where she stayed for two months. During that time her parents failed to visit regularly and learn how to care for her, so she was referred to the Department of Children's Services (DCS).

While in the home, DCS discovered her 11 month old brother Isaiah in horrible condition and he was taken into custody as well. Even though he was not born with MSUD, he had been neglected and was malnourished.

Even though Israelle's MSUD has been a difficult challenge, we look at it as the reason her brother's life was saved. We were originally told that her brain swelling was so severe that she would have suffered brain damage and that we should expect neurological deficits. Over the next two years we fostered our babies, learning and controlling Israelle's condition and dealing with Isaiah's muscular damage and feeding disorder which resulted from

his neglect. The parents visited occasionally but failed to make any efforts to meet the requirements to have them returned to their care, and eventually they stopped appearing in court.

On May 27, 2016 we adopted our sweet babies! We now have 10 children, many of which are handicapped. Each of our children have their challenges and back story. All of them bring such joy to our family. We are so blessed to be their parents!

In the last few years Israelle has progressed so much. She is g-tube dependent and won't take much more than crunchy salty snacks, water, and juice by mouth. She is however incredibly stable. She has only had one serious stomach virus that had her so sick that it worried us, and has been hospitalized only 3 times in 4 years. She is treated at Vanderbilt Children's Hospital in Nashville, Tennessee. Her regular labs are usually perfect! She has surprised everyone by developing without any learning disabilities at all. She is now in kindergarten and has a nurse with her at school. She is very bright, loves to sing, and swims like a fish. Her brother has achieved a healthy weight although he still struggles with a feeding disorder. They are happy and healthy and have completed our family. ■



RESEARCH

MSUD QUALITY OF LIFE SURVEY

By Ed Fischler

The MSUD Family Support Group is teaming with the Clinic for Special Children in Strasburg, PA to conduct a Quality of Life Survey for MSUD families.

There is an urgent need and readiness across patients, drug developers and regulators to understand the critical needs of MSUD patients and caregivers. With this knowledge, clinical endpoints (what researchers should measure when conducting studies) can be identified. The Quality of Life Survey is a crucial missing piece, needed to synthesize these perspectives, resources, and energies to inform therapeutic development and review in this rare and devastating disease.

The objectives of the Quality of Life Survey are to:

1. Understand the many ways in which MSUD affects the quality of life of MSUD families with the aim of developing drug and other treatments which will have the most impact.
2. Identify potential research "end points" or metrics which can be incorporated into the development of MSUD registries, natural history studies, and future research project goals.

The survey will ask patients, parents (or other caregivers), and siblings to rate their experiences of how MSUD affects their day to day life. Questions will focus on:

- The physical effects of MSUD on patients and families, including questions about the MSUD formula,
- The mental stresses caused by the day to day management of MSUD,
- Social interaction issues, and
- The impact of the severity of MSUD on the patient and family member.

We strongly encourage the MSUD community to participate in this one-time survey, but participation is completely voluntary. The survey takes about 30-45 minutes by phone; the survey is administered by the Research Fellow at the Clinic for Special Children, Lauren Bowser. Please contact the Lauren at the Clinic for Special Children at queries@clinicforspecialchildren.org or calling the Clinic at 717-687-9407 and Lauren will return your call and arrange a time to complete the survey over the phone. ■

MSUD RESEARCH: AN OVERVIEW OF CURRENT PROJECTS

2019 Million Dollar Bike Ride Awardees

Our successful fundraising drive resulted in the selection of two projects to advance MSUD research:

"A novel approach for identification of therapeutic leads to the Maple Syrup Urine Disease"

Prof. Ehud Gazit, Tel Aviv University, Israel

Using a yeast model developed for MSUD in their lab, the aim of this study is to understand basic MSUD

pathology with the goal of developing novel treatments.

"Can we improve neurologic and psychiatric outcomes in MSUD? Studies in a new mouse model"

Dr. Rebecca Ahrens-Nicklas, The Children's Hospital of Philadelphia

MSUD typically affects thought processes, mood, and

'Projects' Continued on page 18

GROUNDBREAKING 30-YEAR STUDY IDENTIFIES CRITICAL NEED OF DISEASE-MODIFYING THERAPIES FOR MAPLE SYRUP URINE DISEASE (MSUD)

By Lauren Bowser and Kelly Cullen
Clinic for Special Children

A new study led by the Clinic for Special Children (CSC) analyzed 30 years of patient data and details the clinical course of 184 individuals with genetically diverse forms of Maple Syrup Urine Disease (MSUD). Researchers collected data on survival, hospitalization rates, metabolic crises, liver transplantation, and cognitive outcome. This represents the largest systematic study of MSUD with regard to both cohort size and the duration of clinical follow up. The study was a broad collaborative effort and appears in the journal *Molecular Genetics and Metabolism*.*

Before the CSC's inception in 1989, 1 in 3 children born with MSUD died from neurological complications of the disease before 10 years of age, and the majority of survivors were permanently disabled. Three decades of innovation and clinical care by the CSC team have increased survival from 63% to 95%, while hospitalization rates have decreased from 7 to just 0.25 hospital days per patient per year. Specific advances in management include new prescription formulas for children and adults as well as elective liver transplantation, a collaboration with the Hillman Center for Pediatric Transplantation (UPMC Children's Hospital of Pittsburgh) that has been 100% successful for 93 individuals transplanted since 2003.

Treatment of MSUD requires close monitoring of blood amino acid levels. A total of 13,589 amino acid profiles were generated by CSC's on-site clinical laboratory and the data were analyzed to determine the overall effectiveness of treatment. The authors conclude that although stringent dietary therapy maintains blood amino acid concentrations within acceptable limits, it is challenging to implement, especially for individuals older than 10 years of age, and does not fully prevent the cognitive and psychiatric disabilities caused by MSUD.

Eighty-two (82) MSUD patients underwent IQ testing, with higher IQ scores correlating by age with younger patients. On average, MSUD patients scored 23% lower on IQ testing than their unaffected siblings and, as compared to the general population, the prevalence of affective illness (depression, anxiety, and panic disorder) was much higher among both MSUD patients and their unaffected siblings. Based on these observations, the authors conclude that despite advances in clinical care, MSUD remains a morbid and potentially fatal disorder. There is a critical unmet need for safer and more effective disease-modifying interventions for MSUD.

* Kevin A. Strauss, Vincent J. Carson, Kyle Soltys, et al. Branched-chain α -ketoacid dehydrogenase deficiency (maple syrup urine disease): Treatment, biomarkers, and outcomes. *Molecular Genetics and Metabolism*. 16 January 2020. doi: 10.1016/j.ymgme.2020.01.006 [Epub ahead of print] ■

'Projects' Continued from page 17

behavior. This project will use a mouse model with the aim of better understanding the mechanisms by which this occurs. This knowledge is a necessary step in the development of therapies to protect the brain.

MSUD Family Support Group Supported Research

MSUD Cow Model

Researchers at the University of Massachusetts are working with the Clinic for Special Children to study gene therapy in a cow model of MSUD. The cow model is felt to have advantages over the mouse model used in most research.

Metformin and Rapamycin, Buck Institute

Researchers continue to study the effect of these substances on MSUD in hopes of developing therapies. ■

FOOD AND NUTRITION

LOW-PROTEIN RESOURCES FROM NATIONAL PKU NEWS

By Sarah Chamberlin, Executive Director



It's often said of those of us with rare diseases: "Alone we are rare. Together we are strong." This is definitely true among those of us with metabolic disorders -- each disorder has its own own cause, symptoms, and treatments, but in the end, we have a lot in common. The low-protein diet is a common denominator between those with PKU and those with MSUD.

For more than thirty years, National PKU News has been providing support and resources to people with PKU, but our mandate stretches further than that. Much of our work is designed to benefit the larger community of those with metabolic disorders on a low-protein diet. Here's some information about programs of ours that might be useful to those with MSUD:

- **CookForLove.org** was the brainchild of PKU News board member Brenda Winarski, who refused to believe that a limited diet meant her daughter, born 21 years ago, couldn't love and enjoy food. A speech-language pathologist, Brenda went to culinary school and developed her own delicious low-protein recipe site. In 2018, Cook for Love became a project of PKU News, and was relaunched to include recipes from Brenda and HCU parent & pastry chef Amber Gibson. Additionally, we've begun to add all the recipes in Virginia Schuett's *Low Protein Cookery for PKU*, and *Apples to Zucchini*, co-authored by Schuett and Dorothy Corry. These recipes span decades of delicious low-protein cooking -- from the quick to the complex -- and provide hundreds of options for low-protein cooking, many of them without requiring medical foods. Users can set up a free

account and save their favorites for quick access. Phase 2 of the site relaunch will take place in the coming months, when community members will be able to add and share their recipes with the entire community -- with protein values verified by HowMuchPhe.org. This gives the MSUD community an additional resource to not only find, but share, delicious low-protein recipes. Brenda's Facebook Group - Cook for Love, a Low-Protein Cooking Community is another great resource for support & inspiration for everyone on a low-protein diet.

- **HowMuchPhe.org**: This web app can be a valuable resource for those with MSUD who want to track their protein intake. With unrounded protein values for more than 8000 foods and the ability to chart your intake, it gives you a great picture of your diet & allows you to easily share records with your dietitian/clinical team. We're also expanding the feature set to allow those with metabolic disorders other than PKU to track & chart their blood levels. If you'd like to provide input on what features would be most useful to you as a member of the MSUD community, please contact us at info@howmuchphe.org.
- **Go Low Pro**: In 2018 at the National PKU Alliance conference, PKU News hosted a breakout session in which young adults with PKU, their parents, and adults with PKU were able to discuss their biggest concerns about maintaining the diet in adulthood. Many teens felt unprepared to manage their diet independently; many parents had a hard time letting go of control over their teen's diets, and many adults expressed frustration with having to manage a low-protein diet on top of all their other responsibilities -- jobs, families, and more. Out of this discussion came the idea for **GO LOW PRO**, a low-protein grocery & restaurant locator that could help young adults making new lives away from home at

college, adults traveling for work (or pleasure) and students on foreign study abroad. The idea wasn't a new one: Find Me Gluten Free is a popular crowd-sourced app for those with celiac; Happy Cow does the same for vegans. But the low-protein diet is very specific, and pretty unusual. With support from the Global Genes Foundation and private donors, PKU News launched GO LOW PRO in November 2019. Pre-seeded with 35,000 low-protein friendly stores & restaurants and 75 brands, the app is a great starting place if you're traveling, or in a new area and want to know where to find your favorite products, or a meal that goes beyond the standard salad and fries that many of us settle for. But the key to the app is the users: by creating a profile you can add your reviews & products to the current catalog, enriching the experience for everyone. Now you can find a cauliflower steak in Philly, or the elusive Dole Whip along California's Highway 1, and if you find other delicious options, share them with the rest of the low-protein community.

GO LOW PRO exemplifies the kind of project -- building on our common needs within the metabolic community -- that are most important to PKU News. This has also guided our involvement in advocacy efforts, alongside the MSUD Family Support Group, in support of the Medical Nutrition Equity Act. While many states do have laws that provide some coverage for metabolic formula and low-protein foods, each and every one of them has loopholes that leave many in the metabolic community exposed and without adequate coverage. We're committed to seeing this law passed and thrilled at the collaboration the effort has brought about not only within the metabolic community, but with those with GI disorders who also rely on medical nutrition. If you haven't yet had a chance to do so, please check out [NutritionEquity.org](https://www.nutritionequity.org) and click on "share your story" if you're comfortable doing so: the more voices we can get from the full range of the metabolic community, the stronger our argument for equitable coverage & justice will be. ■

FOOD SAFETY AND NUTRITION DURING THE COVID-19 PANDEMIC

By Karen Dolins, EdD, RD

Most of us are on overload with information from media, friends, and the internet. It is essential that we all stay informed and able to differentiate vital information from hear-say. These websites provide reliable, science-based information:

- **CDC** <https://www.cdc.gov/coronavirus/2019-ncov/>
- **FDA** <https://www.fda.gov/emergency-preparedness-and-response/mcm-issues/coronavirus-disease-2019-covid-19>
- **NIH** <https://www.nih.gov/health-information/coronavirus>
- **WHO** <https://www.who.int/health-topics/coronavirus>

Practice Food Safety

- Wash hands before preparing or eating food. Use soap and scrub for at least 20 seconds.
- Wash hands after coughing, sneezing, blowing your nose, or using the bathroom.
- Clean and disinfect countertops, doorknobs, refrigerator doors, and cabinet doors daily.
- There is currently no evidence to suggest that Covid-19 can be transmitted through food or water systems.
- Avoid sharing food and beverages.
- Refrigerate foods promptly, keeping raw and cooked foods separate.
- Heat food to the appropriate internal temperature.

Access to Food – Advice from the Academy of Nutrition and Dietetics

- During this public health emergency, government agencies have developed flexibilities to help individuals who use programs such as the Special Supplemental Nutrition Program for Women, Infants and Children (WIC) and the Supplemental Nutrition Assistance Program (SNAP). The U.S. Department of Agriculture (USDA) has also developed plans for children who participate in the National School Lunch and School Breakfast Programs so that they are able to have continued access to food during prolonged school closures.
- Older adults and other individuals who are considered at increased risk for complications from COVID-19 should evaluate the foods they have at home. If you are at high-risk or are unable to get the items you need, consider contacting family or friends to assist. Meal delivery and grocery delivery services may be available as an alternative option, and many businesses are offering additional precautions to help reduce the risk of spreading COVID-19.

Supplements and Claims for Cures

- Currently, there are no known cures for COVID-19, though research is underway to develop a vaccine. In its continuing efforts to protect consumers, the U.S. Food and Drug Administration (FDA) has been monitoring and warning companies that offer fraudulent products which claim to help prevent, diagnose, treat or cure COVID-19. Untested supplements and other products touted as a prevention or cure to COVID-19 that are not regulated by the FDA may be dangerous and potentially life threatening.

Staying Healthy with Food

Here are some tips that I'm providing for my clients and that can be applied to those with MSUD and their families.

- Include foods that support the immune system: Nutrients such as protein, vitamin A, D, C, E, and the minerals zinc, copper, and iron are among

those involved in immunity. But nutrition is a team sport and all players are needed to win the game. Eat a wide variety of foods including fruits and vegetables of different color (eating the rainbow) to ensure that you're getting what you need.

- o For those with MSUD, consuming all prescribed formula is critical.
- Eat foods, not supplements: The abundance of research clearly shows that foods, with their combination of nutrients and phytochemicals, are the key to good health and superior to dietary supplements. As noted in a recent report on Natural Remedies and Supplements for Coronavirus by ConsumerLabs.com, supplements will likely benefit only those who are deficient.
 - o MSUD formulas provide all essential vitamins and minerals.
 - o This does not refer to isoleucine, valine, and other dietary supplements that may have been prescribed by your metabolic team.
- The virus is not foodborne. You will therefore not contract it by eating food. Be aware, though, that the virus can be transmitted by touching a food that was touched or breathed upon by someone with the virus and then touching your face. Wash hands before and after touching food. Scrub with soap for 20 seconds before rinsing.
- Wash fresh produce carefully and use soap when possible. Wash oranges and other whole fruit before cutting. Consider soaking items like greens in soapy water and then rinsing thoroughly, especially if you plan to eat them raw. Cooking will destroy the virus.
- Frozen and canned fruits and vegetables are good alternatives. Frozen foods are packed shortly after being picked and retain their nutrients.
- Moderate exercise helps our immune system. Try to include 30-60 minutes of moderate intensity exercise such as a walk or bike ride daily. Exercise classes are also available online. ■



Metabolic Camp

A Holistic Approach to the Management of PKU & MSUD

Established 1995

June 22-27, 2020

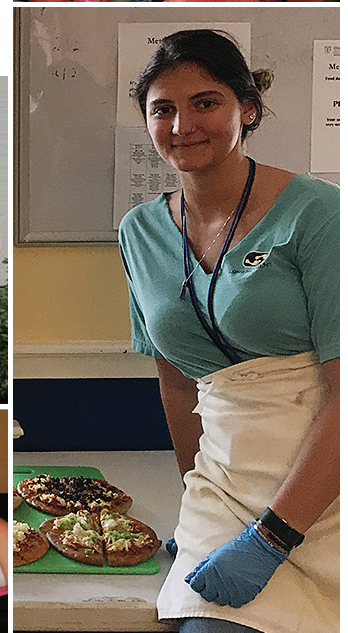
Emory University | Atlanta, GA
Rani H. Singh, PhD, RD, *Camp Director*

metcamp.net



EMORY
UNIVERSITY
SCHOOL OF
MEDICINE

Department of
Human Genetics



Join our annual Metabolic Camp for summer fun, learning, and tools for managing PKU and MSUD!

About

Metabolic Camp offers a holistic approach to the nutrition management of PKU and MSUD—empowering females ages 12 and older to make healthy choices and take charge of their futures as they live, learn, and play in a supportive camp environment. Our model research-based camp, with a focus on Maternal PKU, has been supporting adolescent girls and young women for more than two decades.

Goals

1. To teach the **importance of nutrition** and help the campers develop diet self-management skills.
2. To review the **treatment recommendations of PKU and MSUD** prior to and during pregnancy.
3. To create a place to **share experiences and make new friends** with other young women who have PKU or MSUD.
4. To **research the impact of the above interventions** on the transition to adulthood, pregnancy, quality of life, and overall health outcomes.

Daily Activities

The camp program focuses on building social support through a variety of activities:

- Nutrition education
- Cooking classes
- Discussion groups
- Cooperative sports
- Local field trips
- And more!

The camp will be conducted for five consecutive days. Meals will be provided and supplemented with low-protein modified foods. Registered dietitians will be present to assist at all meals.

Exhibits

Campers and their families are invited to visit exhibits during check-in on Monday, June 22, 2020 at the ATO and MLO Houses at Emory University. The exhibitors will be medical food companies, low-protein modified food companies, and patient support organizations. Families will enjoy a valuable

opportunity to interact with the representatives and to learn more about the products and services offered by these groups.

Accommodations

Metabolic Camp 2020 will be held on the campus of Emory University. Lounge, refrigerator and laundry facilities will be available on site. All participants will be responsible for their own transportation to and from camp.

Faculty

There will be one medical faculty member and one assistant for every 5-7 campers. The faculty will be involved in helping campers with daily activities. In case of any medical emergency, we will use Children's Healthcare of Atlanta at Egleston or Emory University Hospital, where a medical geneticist is always on call.

For more information, please contact:

Rosalynn Blair
Assistant Camp Director
404-778-8521
rborlaz@emory.edu

metcamp.net

Registration

Cost of Camp: **\$375 per person**

Registration Deadline: **March 1, 2020**

Register Online: <http://metcamp.net>

We rely on annual financial support from grants and donations, so we can offer you a subsidized camper registration fee of \$375 per person. This cost includes room, board, and all sponsored group activities. A \$100 nonrefundable deposit is required to hold a place for each camper. This fee will be applied toward the total \$375 camper registration fee. Partial scholarships are available based on financial need. Payment plans and partial scholarships are available.

For more information, please contact:

Rosalynn Blair
Assistant Camp Director
404-778-8521
rborlaz@emory.edu

COVID-19 MEDICAL FOOD ACCESS FACT SHEET

The Emory team has prepared this fact sheet for families and providers. A comprehensive list of medical food companies and parent organizations is provided, along with contact information for any support needed related to medical foods and Low Protein Modified Foods. This Fact Sheet will be updated regularly. A link at the bottom allows you to provide feedback. ■



Medical Food Access Emergency Fact Sheet



Medical Food Manufacturers		
Company	Contact Information	Updated
Abbott	Website: https://abbottnutrition.com/therapeutic#metabolic Consumer Relations: 1-800-227-5767 Mon-Fri, 8:30 AM—5:00 PM EST	March 2020
Ajinomoto Cambrooke	Website: https://www.cambrooke.com/ (Under “Products” tab drop-down menu, select “Metabolic”) General Customer Care & Medical Food Product Support: 866-456-9776 (Option 2) Mon-Fri, 8:30 AM—5:30 PM EST	March 2020
Galen US Medical Nutrition (formerly POA Pharma)	Website: http://www.poapharma.com/en/general-products/ Direct Contact: Ashley Park Phone: 267-382-6416 Email: ashley.park@galen-pharma.com	March 2020
Mead Johnson	Website: https://www.meadjohnson.com/pediatrics/us-en/ (Under “Product Information” drop-down menu, select “Products List”) Patient Care Customer Services: 1-800-BABY-123 (1-800-2229-123) Mon-Fri, 7:00 AM—7:00 PM CST/CDT; Sat: 8:00 AM—4:30 PM CST/CDT Email: www.enfamil.com	March 2020
Medica Nutrition	Website: https://medica-nutrition.com/products/ Phone (USA): 877-850-0985 Phone (Canada): 888-606-6676 Email: orders@medica-nutrition.com	March 2020
Nutricia North America	Website: http://www.medicalfood.com/ (Select “Our Products” tab) Nutricia Customer Service Department: 1-800-605-0410 Mon-Fri, 8:30 AM—5:00 PM EST After Hours: 1-800-365-7354 Mon-Fri, 5:00-PM—7:00 PM EDT	March 2020
VitaFlo USA, LLC	Website: https://www.nestlehealthscience.us/vitaFlo-usa/products Customer Service: 1-888-VITAFLO (1-888-848-2356) Mon-Fri, 9:00—5:00 PM EST	March 2020

Low Protein Modified Food Manufacturers

Company	Contact Information	Updated
Ajinomoto Cambrooke	Website: https://www.cambrooke.com/ (Under “Products” tab drop-down menu, select “Specialty Food—Low Protein”) General Customer Care & Medical Food Product Support: 866-456-9776, Option 2 Mon-Fri, 8:30 AM—5:30 PM EST	March 2020
Dr. Schär's/Mevalia	Website: https://www.mevalia.com/en Customer Service: 0800-988-2488 Email: info@mevalia.com	March 2020
Ener-G Foods	Website: https://www.ener-g.com/ Customer Service: 1-800-331-5222 Mon-Fri, 8:00 AM—5:00 PM (PT) Email: customerservice@ener-g.com	March 2020
Nutricia North America	Website: https://shop.medicalfood.com/category/12/pku-products Nutricia Customer Service Department: 1-800-605-0410 Mon-Fri, 8:30 AM—5:00 PM EST	March 2020
PKU Perspectives	Website: https://www.pkuperspectives.com/ Customer Service: 866-758-3663 Mon-Thurs, 10:00AM—5:30 PM MST	March 2020
National Food Distribution Centre for the Treatment of Hereditary Metabolic Diseases (NFDC)	Website: https://nfdc.info/home.php National Food Distribution Centre: 1-888-606-6676, ext. 221 Email: info@nfdc.info Order: order@nfdc.info	March 2020
Taste Connections, LLC	Website: https://tasteconnections.com/index.php/en/ Direct Contact: Malathy Ramanujam Phone: 310-413-6499	March 2020
Association for Creatine Deficiencies	Website: https://creatineinfo.org/ Facebook Support Group: https://www.facebook.com/creatineinfo Email: info@creatineinfo.org	March 2020
Connecting Families Urea Cycle Disorders (UCD) Foundation	Website: https://ucdfamily.org/ Facebook Support Group: https://www.facebook.com/ucdfamilies/ Phone: 918-490-3055	March 2020
Fatty Acid Oxidation (FOD) Disorders Family Support Group	Website: https://fodsupport.org/ Facebook Support Group: https://www.facebook.com/groups/59945507904/ Direct Contact: Deb Lee Gould, MEd, Director (Family Support Group Parent) Phone: 517-381-5206	March 2020

Patient Organizations		
Company	Contact Information	Updated
Homocystinuria Network (HCU) America	Website: https://hcunetworkamerica.org/ Facebook Support Group: https://www.facebook.com/HCUNetworkAmerica/ Phone: 630-360-2087 Email: info@hcunetworkamerica.org	March 2020
Maple Syrup Urine Disease (MSUD) Family Support Group	Website: https://www.msud-support.org/ Facebook Support Group: https://www.facebook.com/msudfamilysupportgroup/ Direct Contact: Jordan Coleman (Advocacy Chair) Phone: 925-349-4601 Email: coleman.jorann@gmail.com Direct Contact (Espanol): Adriana Carbajel (MSUD Parent) Phone: 480-278-4713 Email: adrianamc2014@yahoo.com	March 2020
National Phenylketonuria (PKU) Alliance	Website: https://www.npkua.org/ Find a Clinic: https://www.npkua.org/Resources/Find-a-Clinic	March 2020
National Organization for Rare Disorders (NORD) Emergency Relief Program	Website: https://rarediseases.org/ Patient Services: 1-800-999-6673 (Espanol: 844-259-7178) Mon-Thurs, 8:30 AM—7:00 PM EST Fri, 8:30 AM—6:00 PM EST	March 2020
Organic Acidemia Association	Website: https://www.oaanews.org/ Facebook Support Group: https://www.facebook.com/groups/33534928222/ Direct Contact: Kathy Stagni, Executive Director (Propionic Acidemia Parent) Email: mkstagni@gmail.com Phone: 763-559-1797 (CT) Direct Contact: Menta Pitre, Director (MMA Cbl C Parent) Phone: 985-856-5631 (CT)	March 2020
Please contact us at semgenetics@emory.edu for any feedback and updates.		

For additional information, please visit www.mnt4p.org and southeastgenetics.org.

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