Staying Safe!

It’s been a year! 2020 can’t end soon enough! I hope everyone is staying safe and healthy. I have been super busy working from home since March. I was asked to share information about OAA at the Metabolic Support UK’s Annual Virtual Conference in October. The breakout session for MMA/PA started at 5:30 AM Central Time on a Sunday morning – thanks to those who woke up early to attend. OAA’s is still planning our face-to-face family conference in Bethesda, MD on June 26/27, 2021. There will also be a Scientific Meeting planned Friday, June 25th hosted by the NIH. More information on the conference can be found on our website oaanews.org/2021-hcuoaapaf-family-conference.html. Erin MacLean is our conference coordinator will be updating the website with the agenda soon. We are hopeful there will be vaccine available and we will be able to see everyone in June!

There are several new treatments for MMA/PA coming to clinical trials in the first half of 2021! OAA is working hard to share the information as we receive it in the newsletter and on social media – giving space to each company. Please talk to your metabolic specialist if you or your child would be interested in participating in these trials.

I’m excited to share that OAA will host a Virtual MMA/PA Town Hall at 10 AM (CST) on December 5, 2020 on Zoom. The team from the NIH will share updated information on the study and research at the NIH. Please check our Facebook Group or Google Group if you’re interested in attending the session.

Kathy

Demolition Derby and OAA/Isovaleric Awareness!

Here’s a few photos of demolition derby cars that were built by brothers Dylan and Cody. The cars ran in the Chaos in the Valley in Winchester, VA In memory of their brother Brandon Tyler Baker who passed away at 28 days old in the year 2000 due to Isovaleric Acidemia. He would have turned 20 years old in October. This one is for him.

Thanks guys for highlighting OAA on your cars!

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Stephanie Carleton and Josh McMahon
OAA Natural History Registry Coordinators
Nika is not one of tribulation but one of triumph. She arrived 2 weeks early, on a Thursday morning, weighing 5.8lbs. There were no complications except she had a low glucose level that was quickly remedied. We marveled at how beautiful and tiny she was. Compared to the other babies, she was by far the quietest.

There were no major incidents that Thursday or Friday except that as a first-time mom, I had trouble nursing. I believed that Nika was not getting enough milk as she go through periods of loud shrieking and hand shaking. I was told that this was normal and that newborns did not need much milk. Her shrieking and “cluster feeding” however only intensified the guilt I felt about my lack of milk. I insisted that she be given formula. I also refused that she be given the Hepatitis B vaccine as I had been reading about the pros and cons of vaccinations and was still unsure of my stance.

We were due to go home on Saturday morning. As Nika was born with low glucose, the nurse checked it once again before letting us go. Her level had declined again. The Nurse urged me to nurse her so that her levels would go back up. With what milk, I wanted to know with frustration. When they returned, they found that her values were even lower. I was sent home and she was kept overnight to get her glucose to normal levels. I remember feeling angry at myself that I did not notice that something was seriously wrong.

Nika’s father and I returned Sunday morning to check her progress. We were told that she had improved. I am not sure what else we may have been told at that time but I remember thinking that she was getting better so she will be home soon. Unfortunately, while sitting outside we received a call that Nika was going to be transferred to Mount Sinai Hospital and that she was suspected of having a metabolic disorder. We were scared since we were unsure of how serious this could be. I just kept saying to myself that God would not have given her to me to take her away so soon.

Nika spent two weeks at Mount Sinai Hospital. We were first told that it may be weeks before Nika was ready to go home as the doctors were unsure of what metabolic disorder she had at the time. A few days later, we learned that they believed she had Propionic Acidemia and were starting a course of treatment. We were also told not to research the disorder on our own because PA patient’s varied and someone else’s story may not be Nika’s. We decided to put our faith in God and the doctors, one of which surprisingly was the daughter of a doctor my grandmother worked with. Nika’s levels improved daily. It was nerve racking keeping up with all the numbers when all we really wanted to know was whether she was better or not. The biggest battle during this period was that she not be sent home with a feeding tube as the doctors were discussing. I knew that she had no problem breast feeding except for my lack of milk so I strongly told the doctors to give her time. At the end of the two weeks, she was taking her goal feeds by bottle and was ready to go home. We were giving emergency protocols, diet plans and numerous other information to care for our special angel.

Nika’s first year was incident free except for a few instances of bad acid reflux that resulted in vomiting. She also had a cold that first winter that lasted a few weeks which also involved frequent incidents of vomiting. One of the biggest challenges during this time was keeping up with the recommended daily protein intake. Nika was not and still is not a large eater. I was so frustrated with reaching the protein goal on a strictly vegetarian diet that I began including small quantities of meat products in her meals. There are also times when we feel pressured to try to get Nika to eat more or to take certain vitamins especially when we hear from the doctors that they would like her to gain more weight. However, we have decided to stick to what we know works. We understand that Nika’s condition makes her more vulnerable but sometimes we feel that the doctors are expecting too much.

Both Nika’s father and I were on the smaller side as children. It is expected that Nika would be slender also. We know she is not starving and the fact that she has been healthy so far attests to that.
As Nika approached her first birthday having experienced no metabolic crisis, the doctors started to talk to us about liver transplants. The transplant would help her body to process more of the propionic acid and therefore lessening the amount of ammonia in her body which held in the 80’s since her discharge. My family and I talked about it, we talked to other PA families and I read what I could find online. Because of the crisis at birth, the doctors were convinced that her version was one of the more serious ones. We knew that she could go years without a crisis or maybe her entire life, but we had to answer the question of whether we were willing to take that risk. What if we waited and she went through a crisis that left her brain damaged? We decided to put her on the donor list.

One year to the day that Nika came home from the hospital and a few days before my BAR exam, we received the call that there was a match. As much as we thought we were ready, I remember thinking that this call could not come at a more ill-prepared time. I told the coordinator that I would call him back. I called family members and my bar mentor to get advice. In the end, we decided to accept the transplant. The surgery was the next morning and lasted about 6 hours. I spent those 6 hours in church asking for prayer. All went well and Nika spent another 2 weeks recuperating. While in ICU, there was a day or two of uncertainty as the medical staff could not figure out why Nika was having a hard time getting her oxygen level to remain at 100% on the ventilator. In the end, after hearing that these machines are not necessarily made for young patients, we decided to take her off and see how it goes. It worked! Nika was breathing on her own. Two weeks later, Nika was again home bound, although we did have the customary battle with the doctors that she was not going home with a feeding tube.

About 1 month after the transplant Nika did have a biopsy as her liver numbers kept fluctuating but the biopsy was normal. The doctors decided it may just be her liver settling in. Since then her numbers have been amazing.

Nika has been slow to develop in some areas. She did not walk until she was 21 months and is slowly learning to speak sentences. It has been a tough year in terms of decision making with the pressures of COVID-19 in terms of minimizing Nika’s exposure while ensuring she develops as normally as possible. We did decide to enroll her in a private day care center in September. She is learning from her peers and we continue to see her progress daily. Nika has also recently qualified for speech and physical therapy through Early Intervention. We pray that these services help her go even further.

Through it all, Nika has been a warrior and an inspiration. She continues to be a blessing to those she meets with her infectious smile. We pray every day that God continues to keep her in his grace.

We would like to say a special thanks to our Family and Friends who have been supporting us in every step of this journey. Also, to the Doctors and Staff at Mount Sinai Genetics Clinic and to the Liver Transplant Center. They have become our second family. Lastly, we cannot forget our OA community for being there whenever we have questions and for the continued love and support.

Dee, Queens, New York
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LogicBio Therapeutics, a biotechnology company based in Lexington, Mass., is advancing research for an investigational therapy for methylmalonic acidemia (MMA). We sat down with Daniel Gruskin, MD, Senior VP of Clinical Development to talk about the status of their MMA clinical program.

Can you tell us about LogicBio and your work in MMA?

LogicBio Therapeutics is a small biopharmaceutical company based near Boston and our leadership team has decades of collective rare disease drug development experience. We are currently focused on developing gene therapies for diseases where it is important to treat patients early in the course of the disease and at younger ages. Our innovative technology, which we call GeneRide, is different than traditional gene therapies in that it is specifically designed to work in growing children and still be durable (last a long time) with one single dose. Our first program is in methylmalonic acidemia, and we are excited to be planning a clinical trial in MMA, the SUNRISE trial, that we expect to start in early 2021. The investigational therapy used in the trial, called hLB-001, is a gene therapy that is designed to merge with the patient’s DNA in their liver cells and use the cells’ natural processes to produce the enzyme that is deficient in MMA.

Who will be eligible for the upcoming SUNRISE trial?

As you know, MMA is a disease which can be caused by mutations (changes in a person’s DNA) in several different genes. Because hLB-001 includes the MUT gene, only those individuals whose MMA is caused by a mutation in the MUT gene will be eligible for this trial.

hLB-001 is intended to intervene as early in the course of the disease as is safe and possible, and as such, we plan to initiate the trial in patients with a confirmed MUT genetic mutation between 3 and 12 years of age. As the trial progresses, we will potentially lower the minimum age of enrollment to as young as 6 months of age in coordination with study investigators and the FDA (US Food and Drug Administration). There are numerous additional inclusion and exclusion criteria for the trial, for instance, anyone who has had a liver or kidney transplant will not be eligible to participate. More information including full trial inclusion and exclusion criteria can be found at clinicaltrials.gov. Decisions about who ultimately is enrolled in the study, including individuals living outside of the United States, are left to the discretion of the study investigators. We encourage any family who is interested in learning more about SUNRISE to speak to their physician as all healthcare decisions including trial participation should be made by you/your child’s healthcare team.

Where will you be conducting the SUNRISE trial?

This Phase 1/2 trial will be running in several specialized centers in the United States with anticipated enrollment starting in early 2021. As trial sites are approved to enroll patients, their specific information will be made available via clinicaltrials.gov. LogicBio will work to cover trial-related costs including travel to eliminate barriers to participation.

We recognize the commitment participants and their families are making and will be as transparent and informative as possible.
Why is the SUNRISE trial focused on younger patients? Will older individuals with MMA be considered at some stage in development of hLB-001?

In MMA, the effects of the disease can start right after birth and can be severe. With early intervention, we hope to potentially minimize the disease effects and prevent or delay further damage from occurring. The age maximum of 12 coincides with when certain co-morbidities such as decreasing kidney function have occurred, so the greatest potential benefit of hLB-001 could come in these early years, which is what we hope to learn from SUNRISE. The results of SUNRISE will inform future development plans including whether we expand the age criteria for future studies.

What else can you tell us about the SUNRISE trial?

While full trial information for participants will be available via trial centers, LogicBio believes that this specific trial design will help provide critical information about hLB-001. The duration of the trial is one year with multiple visits to the study centers (though trial assessments and center visits will decrease over time). There will be only one dose of hLB-001 given which will occur in the hospital setting. One of the keys to studying genetic therapies is to better understand durability (how long a one-time treatment may work in the body). As such, there will be long-term follow up required after the initial 1-year active trial period and that long term follow up will be a critical part of our learnings.

We want to ensure that participating families are fully aware of the trial requirements such as different assessments over time, the drug delivery method, potential risks, and other details, to ensure they are making informed and empowered decisions about enrollment. We recognize the commitment participants and their families are making and will be as transparent and informative as possible. Decisions about trial participation are complex and these decisions should always be made in conjunction with your physician.

What does LogicBio hope to learn from this trial?

This Phase 1/2 trial is an important step in bringing a potential new therapy to individuals living with MMA and we are moving forward with our research in close collaboration with the US FDA (Food and Drug Administration). We have conducted extensive pre-clinical testing for hLB-001 for both safety and efficacy (how well the drug achieves desired outcomes). This initial trial in people will primarily be looking at the safety profile of two different doses in trial participants, and also the effect of the medicine on biochemical markers of MMA and important clinical features as well. We will be closely monitoring participants throughout the one-year trial period and, by doing so, hope to gain a better understanding of safety, tolerability and potential efficacy at each dose. These learnings will help inform our future work towards bringing a treatment to the community.

What can we expect from LogicBio in the future?

Our efforts at this time are largely focused on the successful enrollment and conduct of the SUNRISE trial. The results of this trial will tell us a significant amount of information that will help determine our next steps for hLB-001 research such as future trials. We look forward to keeping the community updated as we have additional information to share.

LogicBio is committed to ongoing collaboration both with physicians involved in the care of individuals living with MMA and the MMA patient community. We are especially grateful to the OAA for their partnership to date and all they have done to help support our work. We firmly believe that by engaging with and learning from you, LogicBio can better serve families affected by MMA. We are eager to provide increased educational opportunities and forums to connect with you in the future.

How can the MMA community learn more about LogicBio and your efforts?

For information about LogicBio and our GeneRide technology platform please feel free to visit our company website at logicbio.com. We are always eager to talk with families; if you are interested in learning about opportunities to connect with us or share feedback, please contact our patient advocacy group at patients@logicbio.com.
Hello! My name is Matthew, I was born in Tucson, Arizona on February 3, 1971. My development was fairly normal. According to my Mom, I met developmental milestones on time, talked early at 9 months but walked late 17 months.

During my infancy when baby food meat was introduced I would vomit, this was an early sign that something was amiss. My pediatrician was not too worried and said just cut out meat, which they did. When I was 10 months old, my Mom went into my room to get me up and I was laying in my crib panting like a dog. We were sent to the hospital by the time we arrived I was in a coma, which lasted about 7 days.

At the time none of the doctors knew what was wrong. The University of Arizona hospital was just opening and had a genetic/metabolic specialist, Dr. Grant Morrow, who after several months of testing diagnosed CblA. During that time all I was allowed to have was sugar water.

Following my diagnosis, I was on a very low protein diet, I remember going to McDonalds when I was little and my Mom ordering me a hamburger and just taking the meat out or pizza and taking the cheese off. I had injections of hydroxocobalamin every day and then once week. Dr. Morrow followed me until he left Tucson when I was 6. Then my pediatrician took care of me, as there were no metabolic specialists in Arizona (until I was an adult). No metabolic crises, if I did get sick with the flu or other virus, we did daily injections and more hydration.

I had a relatively normal childhood, starting playing tennis when I was 6 or 7, did Little League and golf, even wrestled in high school. I was always thin so I was teased a lot growing up, I am still on the lean side. I had some learning challenges, especially with math. I was diagnosed with Attention Deficit Hyperactivity Disorder when I was around 12 and used medication, which was and is so helpful, I continue to take medication today. I attended the University of Arizona and received both an undergraduate and Master’s Degree and I began my career in teaching and coaching high school golf and softball. I met and married my wife Kathy, we have two beautiful daughters, one a student at the U of A and one soon to be 12. My 12-year-old was screened for all metabolic disorders when she was born, thank God, she had none!

After about 10 years I returned to school and got a Masters in Educational Administration, since, I have worked as a Vice Principal and Assistant Director of Exceptional Education for a district here in a Tucson, 71 schools. It is a challenge but very rewarding! As an adult, I eat pretty much a low protein, vegetarian diet, an occasional bit of Mexican food, I work out with weights almost every day and play golf once or twice a week.

Two years ago, I was evaluated at NIH, what an experience! Doctors are very thorough, there was some concern about my kidney function, otherwise pretty healthy, it was recommended that I do injections once a day and continue with my diet. I now have a genetic/metabolic specialist at Phoenix Children’s Hospital, who I see one a year. My Primary Care I began seeing when I was 16 so he has walked the road with me.

All in All, despite my challenges, I am so grateful for the care I had and am receiving, the opportunities and the understanding and support of my family. God bless you all!

Matthew, Tucson, AZ
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Ignacy was born on September 6, 2018 as a completely healthy child. Unfortunately, it quickly turned out that something was wrong. When I think about it now, I can’t believe how lucky we were in those first days. Several seemingly unrelated events saved my son’s life.

Right after birth, Ignacy seemed to be perfectly healthy. We were so happy! The delivery was rather easy and quick, and my baby was sleeping like an angel allowing me to rest. How could I know it was the first symptom of this life-threatening disease? At the time, doctors weren’t worried about it neither, ‘Newborns sleep a lot, that’s normal, he’s fine.’ I’ve heard.

During my pregnancy I was diagnosed with gestational diabetes. My results were borderline normal and I only had to watch my diet, but due to that Ignacy had blood tests done right after delivery. This is how we know that everything was still fine then.

On the second day those tests were repeated because Ignacy lost 12% of his weight. This time his blood gases weren’t that good. I remember talking to the doctor at that point. The doctor reassured me that he might have got a little dehydrated and after the drip everything should be ok. Today I know that a moment later she made a call to her boss - my mom.

It just so happens that my mom has been working as a neonatologist for over 30 years, and since 2013 she has been the head of the Neonatal Ward with the Neonatal Intensive Care Unit at the hospital where Ignacy was born. In her career, she saved many children, but she never thought that she would have to fight for her grandson’s life.

How lucky we were that the doctor on duty made that call! It was late in the evening and my mom was already home. The blood gas analysis showed metabolic acidosis. The symptoms presented by Ignacy - reluctance to drink, apathy - are usually considered sepsis and making such a diagnosis would have led the treatment in this direction. But, for some reason, my mother thought they should check his ammonia level first. Even our metabolic doctor said he wouldn’t think of hyperammonaemia at that point.

In many Polish hospitals, it is not even possible to determine ammonia. In the hospital where Ignacy was born, that test was introduced only a year earlier! The blood gas apparatus used at NICU was obtained by my mother thanks to a grant from Norwegian funds just two years earlier. Easy access to these tests certainly helped to make such a quick diagnosis.

Today I can say with certainty - Ignacy lives thanks to his grandmother. Had it not been for her vigilance and insight, this story could have been shorter. Ignacy’s ammonia level was indeed elevated! It decreased after a few hours when the protein intake was stopped and intravenous glucose was started, but they kept it monitored. Thanks to that, Ignacy did not fall into a coma, nor did he need dialysis.

The next day, my mom was on duty at NICU. As ammonia levels rose again, she called a metabolic consultant who was just getting off the plane, returning from a conference in Greece. Only he could help us, because he had access to ammonia-lowering drugs and knew what to do. He came to our hospital at 1 am and together with my mom started a rescue action. I don’t want to think what would have happened if Ignacy had been born the day before, when the doctor was still in Greece. He saved my baby’s life that night.

Thanks to these coincidences, and most of all thanks to the great commitment of the doctors, Ignacy had a pretty good start considering the diagnosis. On the 5th day we knew it was Methylmalonic acidemia, but it took us over a year to determine the specific type (CblB).

Methylmalonic acidemia is a very rare disease, only a few children live with it in Poland. The first hospitalization after diagnosis lasted a month and we were so terrified when we left the hospital. We didn’t know what to expect. Fortunately, I found the Organic Acidemia Association website. Thanks to this and other support groups, I met parents of children like Ignacy. It gave us a lot of strength and hope for the future.

We were also able to connect with specialists from other countries, who helped us a lot and gave us a valuable guidance. Ignacy turned two in September. During those two years, he was hospitalized many times. When he was one he had an intravenous port and a G-tube placed. Unfortunately, as a complication of MMA, he developed recurrent pancreatitis.

Outside the hospital, however, we try to live a ‘normal’ life. Ignacy is a very active and happy boy. He loves running, riding his bike and playing in the pool. We have learned to live in the moment and enjoy every, small or large progress he makes.

Best wishes from Poland!
Ignacy, Ola and Bartek
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CoA Therapeutics to Develop Potential New Treatment for MMA/PA

CoA Therapeutics was founded in 2018 with the aim of developing new therapies for difficult to treat and rare genetic disorders by harnessing advances in understanding of the coenzyme A pathway. We are currently developing an oral, small molecule activator of an enzyme involved in coenzyme A production and we believe that increasing coenzyme A may address some aspects of impaired metabolism in organic acidemias.

CoA Therapeutics' approach is based on breakthrough scientific developments led by Suzanne Jackowski, Charles Rock, Richard Lee and Stephen White at St. Jude Children’s Research Hospital in Memphis, Tennessee. The CoA Therapeutics team is led by a veteran biotechnology team whose sole objective is to partner with patients and physicians to bring effective therapies to market at the soonest possible opportunity. We have completed key efficacy and safety studies in animals and are currently interacting with FDA with the goal of starting our first human trials in 2021.

We are committed to the organic acidemia community and will keep you up to date on new developments. We will continue to work as quickly and as diligently as possible to support patients. If you would like further information or have any questions, please do not hesitate to contact us at info@coatherapeutics.com.

CoA Therapeutics is a member of the BridgeBio family. BridgeBio was founded in 2015 to create treatments for rare diseases and conditions which are caused by a genetic defect. BridgeBio is currently developing treatments for over 20 rare genetic conditions. Some of the treatment programs are at a very early stage, others are in trials, and a small number will be reviewed by the FDA in the next months in preparation for general use.

Helpful links: coatherapeutics.com/, https://bridgebio.com

Martin
Propionic Acidemia
Age 5 months

Our son, Martin, was born on 05/25/2020, well and healthy, we were discharged on 05/27, and we observed on 05/29 loss of appetite and altered breathing, on 05/30 following with these symptoms and also low temperature. We talked to the doctor Pediatrician and went to seek emergency care, there they provided the first care and we were referred to the ICU, due to the signs and symptoms we had two suspicions, a sepsis or an inborn error of the metabolism. They began to treat the most common situation, sepsis, and the next day there was no improvement. We were transferred to the state reference hospital in inborn errors, and we achieved a rapid diagnosis. We were hospitalized on Saturday, Sunday there was the transfer to the reference hospital and Monday we received the diagnosis of Propionic Acidemia.

We were hospitalized in the ICU for 24 days, Martin was metabolically stabilized, but other neurological and hematological issues arose, but he left without any consequences of this situation.

He’s a beautiful, active, very smart baby. A great warrior! We parents and the genetics medical team are very optimistic about Martin’s development.

In a very brief way this is our story so far. We went through a horrible moment of great anguish and fear, a situation we never imagined going through and feelings we had never felt before.... but thank God we’re okay!

(Translated from Google translate – apologize for any errors)

Priscila, Rio Grande do Sul Brazil
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Ammonia Monitoring from a single drop of blood

Thomas Veltman, Ph.D.
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In an earlier article in this newsletter, I described my efforts, along with a team of other researchers, towards producing a point-of-care ammonia meter with functionality similar to the well-known blood glucose meter. In June, we published a cover article in ACS Sensors (doi:10.1021/acssensors.oc00480) showcasing this technology and the performance of a prototype meter and consumable cartridges. The technology demonstrated excellent agreement with the existing plasma ammonia test performed on real patients at Lucille Packard Children’s Hospital and Stanford Hospital. The cartridges enable rapid (~1 minute) and accurate ammonia measurement from a single drop of blood with values between 10 and 1000 μM.

Spurred by the success of our research, we founded Aza Technology to commercialize the technology and hired CEO Peter Karkantis. Peter is an Abbott Laboratories veteran executive with extensive experience in point-of-care glucose testing. Thanks to the support of the OAA, Aza Technology was able to begin work on developing a cartridge design for manufacturability. We have recently constructed a system for producing cartridge components in the thousands of units. In parallel, we have designed and built a feature-complete meter and have begun providing units to potential users for evaluation and feedback. Our new instrument and cartridges are designed to be manufacturable on scale, allowing us to reach the patients that need it most. We are currently developing strategies for FDA evaluation, and seeking partners and funding for full commercialization.

2021 OAA CALENDAR
createphotocalendars.com/shop/organicacidemiasassociation

Thanks to Raymonde DeGrace for creating our fabulous calendar!
Other items are still available at our CafePress shop
www.cafepress.com/organicacidemiasassociation
Propionic acidemia (PA) is a severe life-threatening disease for which there is currently no truly effective treatment. The disease is caused by mutation in one of the two genes that code for the enzyme propionyl-CoA carboxylase (PCC). This enzyme is made up of two different proteins that fold around each other into a complex structure with six of each of these two molecules. This complicated structure and folding pattern make classical gene therapy a very difficult proposition for PA. Recent work in our laboratory has found that a number of specific mutations that cause PA cause problems by interfering with the protein folding and/or assembly process leading to a non-functional enzyme and thus the disease. In cells, proteins with complicated folding patterns are often helped to fold by other proteins called chaperones that guide the protein down the correct folding pathway towards the final functional assembled protein. We have observed that a number of mutant forms of PCC can be restored to normal activity if they are helped to fold correctly using these chaperone proteins. In our current study, we will examine a number of chemicals that can also function as chaperones and assist with protein folding with a view towards restoring full activity in mutant forms of PCC. This work will initially occur in a bacterial PCC expression system as a proof of principle to identify promising compounds and then depending upon progress, move into treating human PCC patient derived cells. In the long term, we would envisage screening huge libraries of different chemical compounds with a view towards identifying future drugs that are both effective and safe for PA and targeted towards specific mutations in PCC. Our current studies have the potential to serve as an initial first step in the rational design of a personalized medicine strategy for patients with specific mutations causing PA and we gratefully acknowledge the financial support of the Propionic Acidemia Foundation in this endeavor.

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Hi -- we have officially opened up the website to order a Rare Bear. This is our fourth year partnering with Rare Science. There is no charge for the Bears -- OAA assists with the funding for the postage. Please do not request another bear if you received one in the past three years or requested one through another organization. You will need to complete the form entirely and click “Submit” at the end -- a CAPTCHA is required to eliminate any spam...so be sure you click on that too.

Rare Science has a ship date for our group - December 12th!! They will be closing registration for new bears on November 30th - so if you haven’t signed up - do so before the end of the month!

There is a good chance the bears will arrive by Christmas (probably not the International addresses). Here’s the link to register - please be sure to fill out completely or it may not be processed.

www.rarescience.org/oaa/
Online MMA and PA Study Recruiting Participants

HemoShear is now recruiting participants for the MMA and PA Insight Study. This pilot study will evaluate how different assessments can be used for people with methylmalonic acidemia (MMA) and propionic acidemia (PA) to measure verbal communication, processing speed, life skills, quality of life and sleep.

A pilot study is a research study with a small number of participants that takes place before a planned larger study to make sure that the methods of the larger study will be appropriate. This pilot is designed to identify which assessments might show changes in thinking or behavior when a potential new treatment is studied.

“Sometime measuring an improvement can be hard in a clinical trial. I think that this study will help us make the right choices about measures so that future therapeutic studies can demonstrate benefit when they have benefit,” said Kimberly Chapman, MD, PhD, an expert in the diagnosis and management of MMA and PA at Children’s National Hospital in Washington, DC.

Better Testing Needed

Many children and adults with MMA and PA have difficulties learning, communicating, relating to others and participating in activities of daily living. As new treatments for MMA and PA enter into clinical studies, researchers will need effective ways to measure improvements in processing information, communicating and functioning in general.

The MMA and PA Insight Study is designed to identify the most useful tests for clinical studies and determine whether it is possible to conduct testing online with children and adults so this approach can be used in future research studies.

Participants Needed

HemoShear is seeking at least 25 subjects with MMA or PA to join the study. Participants need to be 3 years of age or older, with or without having an organ transplant. Parents/caregivers of children or adults who are unable to participate will be asked to complete the questionnaires.

To learn more, visit MMA-PA-Insight.com

Participation is From Home for Everyone

This study is being conducted online and does not involve any visits to a clinic. Participants will take a few tests online and have a video call with a study investigator.

The testing session will include a brief background interview with the MMA/PA participant or caregiver, if the child is too young or the adult is unable to participate.

- Children 3 to 6 years of age will not participate directly, but their parents/caregivers will complete several questionnaires.
- Children 6 years and older will take one or two brief tests for a total of about 30 minutes.
- Adults with MMA or PA will take two tests for a total of 30 minutes.
- Parents/caregivers will complete a 30-minute online questionnaire and join a video interview with an investigator for about 20 minutes.

Why Participate?

Volunteers for this study will be given a summary of the subject's performance on the tests. You can feel good that the results of the study may help to understand the effects of MMA and PA and the ways to measure how well new potential treatments work in clinical trials. Families will receive a gift card to recognize their time and effort.

To learn more and sign up to be contacted, visit MMA-PA-Insight.com
THE ORGANIC ACIDEMIA ASSOCIATION PRESENTS:

MMA/PA VIRTUAL TOWN HALL

Saturday, December 5, 2020
10:00 AM (CT) 11:00AM (ET)
Zoom Webinar - Registration Required

STAFF FROM THE NATIONAL INSTITUTE OF HEALTH - NIH
DR. CHARLES VENDITTI
DR. OLEG SHCHELOCHKOV
DR. IRINI MANOLI
A new study summarizes over 30 years of clinical experience in the treatment and management of glutaric acidemia type 1 (GA1), a rare and potentially devastating metabolic disorder caused by variants in the GCDH gene. The study followed the clinical course of 168 individuals with GA1 who were born between 1973 and 2019 and originated from 26 states and 6 countries. Participants were divided into three cohorts based on timing of diagnosis and method of treatment. The study was a broad collaborative effort led by clinicians and researchers at the Clinic for Special Children (CSC) and will appear in Molecular Genetics and Metabolism. It establishes a safe and highly effective standard-of-care for the treatment of GA1, and should serve as a rich and valuable resource for dieticians, physicians, and GA1 families throughout the world for years to come.

Before the CSC’s founding in 1989, 90% of infants and young children with GA1 suffered a catastrophic form of acute neurological degeneration. The brain injury of GA1 leaves children mute, wheelchair-dependent, and fully disabled by generalized dystonia, and often results in complications such as scoliosis, hip dislocation, pulmonary aspiration, chronic pain, and untimely death.

Today, with the benefit of early diagnosis, dietary therapy, and an effective hospital protocol, only 7% of children born with GA1 suffer brain injury. Specifically, state newborn screening coupled with strict dietary management reduces the risk of brain injury 14-fold, and uninjured children with GA1 have normal growth, motor development, and cognitive function. Overall, early diagnosis of GA1 with lysine-free, arginine-enriched metabolic formula and emergency IV infusions during the first two years of life is safe and effective – preventing over 90% of brain injuries. The need for dietary and emergency IV therapies beyond early childhood is uncertain at this time.

The research was conducted by a team including the study’s first author Kevin A. Strauss from the Clinic for Special Children, Strasburg, PA; Department of Pediatrics, Penn Medicine – Lancaster General Hospital, Lancaster, PA; and Departments of Pediatrics and Molecular, Cell & Cancer Biology, University of Massachusetts School of Medicine, Worcester, MA; senior author D. Holmes Morton from the Clinic for Special Children, Strasburg, PA; Department of Pediatrics, Penn Medicine – Lancaster General Hospital, Lancaster, PA; and Central Pennsylvania Clinic, Bellefonte, PA; Katie B. Williams, Lauren E. Bowser, Millie Young, Donna L. Robinson, Christine Hendrickson, Keturah Beller, Erik G. Puffenberger, Karilla W. Brigatti from the Clinic for Special Children, Strasburg, PA; Vincent J. Carson and Laura Poskitt from the Clinic for Special Children, Strasburg, PA; and Department of Pediatrics, Penn Medicine – Lancaster General Hospital, Lancaster, PA; Cora M. Taylor and Barbara Haas-Givler from the Geisinger Autism & Developmental Medicine Institute, Lewisburg, PA; Jennifer Hailey from Wellspan Philhaven, Mount Gretna, PA; Stephanie Chopko from the Department of Pediatrics, Nemours/Alfred I. duPont Hospital for Children, Wilmington, DE; and Freeman Miller from the Department of Orthopedic Surgery, Nemours/ A.I. duPont Hospital for Children, Wilmington, DE.

This study was funded in part by a research grant from Nutricia North America, the manufacturer of Glutarade Junior and Glutarade Essential metabolic formula. No author received direct or indirect personal compensation from Nutricia or owns shares in the company. There are no other actual or potential conflicts of interest to declare. About the Clinic for Special Children

The Clinic for Special Children (CSC) is a non-profit organization located in Strasburg, PA, which provides primary pediatric care and advanced laboratory services to those who live with genetic or other complex medical disorders. Founded in 1989, the organization provides services to over 1,050 individuals and is recognized as a world leader in translational and precision medicine. The organization is primarily supported through community fundraising events and donations. For more information, please visit www.ClinicforSpecialChildren.org
In Memoriam

Stacey, Propionic Acidemia
August 5, 1973 – August 26, 2020

Stacey Christine passed away suddenly at home in Red Deer, Alberta, Canada on August 26, 2020 at the age of 47.

Stacey was born in Edmonton, AB on August 5, 1973 to parents Wendy and Don.

She was very ill since birth with a rare metabolic illness, propionic acidemia. She defied all odds and was one of the oldest living persons with this catastrophic illness.

She attended school at Penhold, Innisfail and River Glen. She also attended the trans vocational program and child care program at RDC, for which she was very proud of her accomplished.

She married Kevin John in 1997 and bought their first home in Red Deer. Defying all odds again, Nicholas James was born on January 12, 2000 and Emily Mackenzie on October 14, 2003. Stacey was an amazing, dedicated and loving mother and her children were her true accomplishments in life.

Stubborn, determined, feisty, funny, courageous, inspiring, resilient, strong, thoughtful and loving.

Stacey wrote to over 50 pen pals all over the world and always remembered the special days for family and friends with cards and letters and encouraging words.

Enormous thanks to Dr. Alison Chan, genetics department, UAH, Edmonton, Dr. Ernest McCoy her childhood savior, the doctors and nurses and staff at the RDRH Emergency, ICU and units that have been so kind and concerned about her.
Organic Acidemia Association

(OAA) provides information and support to parents and professionals dealing with a set of inborn errors of metabolism collectively called organic acidemias. The OAA is a volunteer organization registered with the IRS as a 501(c)(3) non-profit corporation. Donations to the OAA are tax deductible. OAA publishes a newsletter three times a year, hosts a Google Group for information exchange and maintains a website and Facebook page. Services are funded by corporate and individual donations. Annual membership donation of $25 (US) and $35 (international) plus $5 for the family roster is requested, but not required. Our 501(c)(3) non-profit status qualifies OAA for United Way donations through their write-in option. If there is a write-in option, just write “Organic Acidemia Association” in the blank line on your pledge card.

Donations can also be made at OAA’s website through the “PayPal” and the “Network for Good” option.

- The information contained herein does not necessarily represent the opinions of our Board of Medical Advisors or Board of Directors
- Letters and photographs sent to OAA become the property of OAA and may be used or edited at the discretion of the OAA staff.
- Names or information will be kept confidential only if specifically requested in writing
- This newsletter does not provide medical advice. You should notify your health care provider before making treatment changes.

ANNUAL DONATION / CHANGE OF ADDRESS
Please accept $___________ as our annual tax deductible donation to the Organic Acidemia Association.

Suggested membership donation is $25 (US) and $35 (international). Extra funds are welcome and can be designated for research, OAA operating expenses, or to help others attend conferences.

Remember the newsletter does not get forwarded when you move!

Name: _______________________________________
Address: _____________________________________
City State Zip: _________________________________
Email: _________________________________________

Mail to: OAA
9040 Duluth Street
Golden Valley MN 55427