Summer/Fall 2022

Hope you all have enjoyed the summer of 2022. It seems to always go by so fast! We started the summer out busy with conference planning! We were so happy to host our in-person conference this past June in Bethesda, MD. We have not been able to gather since 2018. We partnered with the HCU Network America and PAF Support groups this year. I’ve included a few photos in this issue from our time in Bethesda.

We professionally videotaped each speaker session – the presentations and videos can be found on our new and improved OAA website! I’m happy to report that our new OAA website should be ‘live’ by the time you are reading this newsletter! We are strengthening the voice of community by providing an easier to navigate website. We have restructured the content for ease of use and added a new “Blog” that will highlight the stories from this newsletter as well as other new and relevant “breaking news” in between the newsletters. We hope you find the new website easy to navigate, allowing you to get to the information you are looking for quickly. If you are interested in sharing your story for our next issue of the newsletter, please let me know!

Kathy
## Organic Acidemia Association

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OAA GRADUATES
YEAR 2022

Ava - MMA CblC

2021
Ayden - GA1

Zach - GA1

Ayla - MMA Mut 0

Tiffany - PA

Taryn - MMA CblA

Raechel - MMA CblC

Carson - PA

College Academy
Homeschool
Pre-K
Grade 8
University
Kindergarten

OAA GRADS 2022
Organic Acidemia Association

Congratulations
High School
CblC Onlus, HCU Network America and the Organic Acidemia Association today announced the recipient of their first collaborative cobalamin research grant—awarding a research grant to the National Research Council’s Institute of Biophysics in Palermo, Italy to identify potential treatment for cobalamin C (cblC) deficiency. The research, led by Dr. Silvia Vilasi, aims to identify compounds that could potentially rescue MMACHC functional deficiency in cblC disease. Dr. Vilasi is a researcher at the Institute of Biophysics (IBF) in the National Research Council, and has had a longstanding interest and involvement in the study of structure-function relationship of proteins involved in human pathologies. More recently she focused her interest in homocystinuria research.

Combined methylmalonic acidemia and hyperhomocysteinemia – cobalamin C subtype, more commonly known as cobalamin C (cblC) is a rare autosomal recessive metabolic genetic disorder. CblC disease is caused by more than sixty mutations in the gene coding for the MMACHC protein that transports and chemically transforms B12. Patients often present in infancy with megaloblastic anemia, lethargy, growth and developmental delays, cognitive impairment, seizures and progressive retinal deterioration. Late onset cblC often presents with neuropathy or mental health disturbances.

The current treatment consists of hydroxocobalamin injections (a very specific form of B12) and betaine anhydrous. If a patient is symptomatic at the time of diagnosis, treatment will not reverse the clinical sequelae. Despite current treatment standards, many patients still become symptomatic. The exact incidence is unknown and varies globally. It is estimated that cobalamin C impacts at least 1 in 100,000 people worldwide with Italy being one of the countries with the highest known incidence. It has been given the classification of rare disease by the US Office of Diseases Research and is included as part of the newborn screening panels in many countries.

According to the principal investigator, Dr. Silvia Vilasi, the project aims to characterize and classify some of the most common cblC variants of MMACHC protein based on the impact that specific mutation has on MMACHC molecular features, such as structure, stability, B12 binding properties and function. Based on this classification, MMACHC-specific molecules with potential therapeutic benefit and safety will be screened exploiting a structure-based bioinformation approach. The idea is to find molecules able to bind the mutants and stabilize these protein variants in a conformation similar to the unmutated protein (wild type), recovering their function. The protein druggable sites will be targeted with several virtual libraries of drug-like molecules, giving priority to DrugBank library that includes EMA and FDA-approved molecules. 5-10 compounds from the bioinformatic experiments will be then experimentally validated, assessing their ability to restore functionality of proteins. The selected molecules could be precursors for further preclinical studies. Dr. Silvia Vilasi says “I am very happy and honored to have the possibility to contribute to homocystinuria research and I am grateful to CblC Onlus, HCU Network America and the Organic Acidemia Association for the trust they have placed in the project. The team I will coordinate with at IBF can leverage a wealth of facilities, multidisciplinary skills and backgrounds of the members participating in the project, from experimental biophysics, chemistry, cell biology, structural biology and structure-based drug screening. Moreover, to achieve the proposed goals we will collaborate with Prof. Carlo Dionisi Vici, Head of Clinical and Research Unit of Metabolic Diseases at the Ospedale Pediatrico Bambino Gesù in Rome, who will have an important role in mentoring and guiding the experimental activities as the project is designed to be ‘patient-centered’.”.

Rossella Brindisi, President of CblC Onlus says, “It is a great pleasure to collaborate with HCU Network America and Organic Acidemia Association on this project. It is a good example of cooperation among different organizations spread out in different countries to support scientific research and family community. We hope it will pave the way for further common initiatives.”

President of HCU Network America, Margie McGlynn says, “While HCU Network America’s prior grants were awarded for research projects for potential new therapies for classical homocystinuria, we are pleased to collaborate with cblC Onlus and the Organic Acidemia Association to support a grant focused on potential new therapies for cobalamin C disorder, which is consistent with the expanded focus of HCU Network America. We also hope this project will [continued next page]
generate insights that can be applied in the future to other cobalamin disorders.”

Organic Acidemia Executive Director, Kathy Stagni says, “The Organic Acidemia Association is happy to collaborate with HCU America and CblC Onlus on this grant request. We feel fortunate and hopeful that researchers work toward the goal of better treatments for our cobalamin C families.”

HCU Network America, the Organic Acidemia Association and CblC Onlus thank their community of supporters whose contributions made this grant possible.

About CblC Onlus

The CblC Onlus, the Italian Association of Methylmalonic Acidemia with Homocystinuria cblC type, was set up in March 2017, by the initiative of some parents of children affected by such disorder, with the purposeful intention to support scientific research and every initiative aimed at improving the quality of life of patients and their families.

Currently close to 80 families are associated with cblC Onlus.

About HCU Network America

HCU Network America is a 501c(3) non-profit organization founded in 2016 dedicated to helping patients and their families affected by Homocystinuria (HCU) and related disorders. The mission of the organization is to inform and provide resources for patients and families, create connections, influence state and federal policy, and support advancement of diagnosis and treatment for HCU and related disorders.

About the Organic Acidemia Association

The Organic Acidemia Association is a 501c(3) non-profit organization whose mission is to empower families and health care professionals with knowledge in organic acidemia metabolic disorders. We support early intervention through expanded newborn screening, solicit contributions and distribute funding that supports research toward improved treatment and eventual cures in the areas of Organic Acid disorders.

Henry

MMA, Cobalamin C

Age 2

Henry was born in 2020 in Missouri. I had a normal pregnancy and delivery. He was born at 39 weeks, but only weighed 5lbs 13oz. This was my first clue that something was up. We struggled to keep his sugars where they needed to be after delivery, but they assured me this was common in the low weight babies and not to worry. We went home a couple of days later. The day after we got home, I got a call from a genetic counselor telling me Henry had markers show up on his newborn screening. They couldn’t tell me if anything was definitely wrong or not, but they did say that if something was wrong, it could potentially be very serious. They told me to feed him every 3 hours, and if he started to not tolerate his feeds then to take him to an ER. The only thing I noticed was he did seem to sleep a lot, but he was a newborn so I didn’t think too much of it. We ended up having blood draw and starting an oral med while we waited for some results. We also ended up doing an injection 1-2 times a week until we got the results back, which seemed to take forever. We finally were told that it was cblC and that Henry would have to eat on a strict schedule and that he would have to take lots of meds including a daily shot that we were trained to do.

This was an incredibly difficult time. Waiting was the worst, not knowing what was wrong, let alone how to fix it and having this TINY baby to take care of. Then having to go through all the blood draws and having to do shots, it’s so hard to watch your little one go through all that. Being the one to actually give the shots was hard too, i was the one hurting him. Once we got him on all the meds he needed, we finally were able to start trying to settle in and form a routine. Henry grew well and met all of his milestones. He is a ridiculously happy child who is absolutely bursting with personality. He’s always been very alert, even as a newborn, when he was awake he was very focused on you. He runs around with his older siblings all the time and is very bossy with them. Being outside is definitely his favorite. So far Henry has been asymptomatic, with the meds his levels are well controlled. He’s a little delayed in his speech, but it’s hard to say if that’s due to his disease or just because he’s the baby and we all cater him. Having a child with a rare disease is ridiculously hard. The unknowns, the worries, the “what ifs”, the treatments and schedules, the appointments and lab draws, and medical bills. It’s a lot, and it’s hard to watch him go through it all. But it is also so much more than worth it. Henry is thriving and he enjoys life every day, even on the hard ones. We are so grateful God gave us this beautiful boy.

Clarissa
Missouri
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In the fall of 2014, we were living the “classic” American dream. We had a 2-year-old daughter, a yellow Labrador and were about to have our son. Mason was born December 5th, 2014 in Evanston, Wyoming and everything was going “normal”. He had a little bit of nystagmus and was a little tired after his circumcision, but the doctors and nurses said everything was fine and to go home and enjoy your healthy baby boy. Little did we know we were about to become fully aware of the world of rare metabolic diseases.

On Mason’s 6th day of life, we were just hanging out at home and enjoying our new addition to the family when we got a call from the Children’s Hospital in Colorado. They had the results of Mason’s newborn blood screening and his levels were not where they should be. They said that it is very likely that he has a metabolic disease and they recommended we get a hold of his pediatrician to get additional labs done that day to know the next step we need to take. The visit with the pediatrician was a whirlwind. They were fairly confident he had a metabolic disease but didn’t really know which one. They informed us of a few worst-case scenarios to attempt to mentally prepare us for the worst. When we left the doctor’s office, we sat in the car trying to figure out what to do next. We were still overwhelmed by the whirlwind of information we received and the unknowns of which metabolic disease and associated complications he may have. As we sat there Shandra was trying to feed Mason and he wouldn’t latch on. The next thing we knew he was as limp as a noodle, completely lethargic. We marched right back into the doctor’s office for help. Calls were made to the Children’s Hospital and as a group we decided the best thing to do was get Mason to Colorado. In a rushed panic we got our belongings together and had friends take care of our house and our dog. Shandra jumped in the life flight airplane with Mason, and Brandon took Madyson and made the 6-hour drive in the dark in December to Denver. Looking back we did have one thing on our side. The weather and roads were in amazing shape for that time of year.

We spent the next 9 days in the NICU at Children’s learning more than we ever expected to know about Cobalamin C deficiency. It was a lot to take in, but they were able to break down Mason’s condition into terms that we could understand. They ran several tests to understand his current metabolic state as well as a thorough examination of his eyes and his heart. Cobalamin C has several unpredictable side effects, but one that is common with Cobalamin C deficiency is vision loss. During this initial visit Mason did not show any signs of vision loss.

We met some amazing doctors and nurses at Children’s and everyone took great care of us. They helped us establish contacts with all the pharmacies for Mason’s unique meds, which is quite an ordeal for the cystadane and compounded hydroxocobalamin. They also made sure that we were comfortable giving Mason his shot every day. In all openness it was an intimidating task, but we knew he needed it to live so we cowboy’d up and learned how to do it. After his levels were in reasonable shape and they were comfortable that he had all his meds they let us go home.

Once we got to our home in Green River, WY we had to figure out our new complex life. Keeping a watchful eye on our son for potential seizures, lethargy, and the other issues the doctors had warned us about. We were very paranoid about missing one of Mason’s critical doses of his medicine so we built a checklist to make sure we didn’t miss any doses. Figuring out the best schedule to get Mason to the local hospital so that he can get his blood pulled for lab work every week. Reflecting on this we feel we did tremendously well. Mason is now 7 years old and he hasn’t missed a single dose of his life critical medicines.

Since we were so far from the Children’s Hospital we couldn’t visit them every week per the normal protocol, so instead we would go down once a month. Mason’s first visit back to Children’s would be one that we would never forget. At the end of his eye exam Dr. McCourt told us that she could start to see the center of his retinas start to die. Atrophy and the beginning stages of macular degeneration. Dr. McCourt was amazing through this traumatic time. Explaining what she was seeing in terms we could understand. Referring us to different resources and most importantly giving us time to deal with this news. We spent the next 45 minutes sitting in the exam room crying our eyes out. Once we gathered ourselves, we thanked Dr. McCourt for her kindness and we headed out.

We continued to make our monthly trips to Colorado so that the doctors could continue to monitor Mason. At 6, 12, and 18 months we had longer visits where Mason was sedated and they performed MRIs for monitoring. Through the checkups Mason continued to have high chemistry for a normal individual, but they were on the lower side that the doctors had seen for a Cobalamin C patient. It felt good that the hard work and diligence to making sure he got all of his medications was paying off. The doctors were impressed...

[continued next page]
and completely baffled that the dead spots on his retinas was continuing to grow with good metabolic chemistry. Dr. Weisfeld-Adams (metabolic) and Dr. McCourt (ophthalmology) were determined to figure out what was causing the vision loss and we promised to help them out in any way that we could. As the visits continued, Mason’s vision has been relatively stable since he was about two and a half years old. Dr. McCourt continues to closely monitor to make sure that they dead spots in the center of his eyes do not grow and closely monitors his peripheral vision for any signs of loss. Unfortunately cancer took Dr. Weisfeld-Adams away from us in 2018. We were heartbroken that he is no longer here, but we still have amazing metabolic doctors that take care of Mason. We were honored to be invited to his funeral. Brandon was able to speak during his services and let his close family and friends know how special he was to our family and how he was able to make this extremely difficult situation we were living with endurable.

As Mason has grown up, we have done our best to let him try anything and never hold him back. He has played indoor soccer and participated in the local baseball “Challenger League”. Soccer is getting a little tougher for him as the kids are getting faster and more aggressive. He has fallen after bumping into the kids and he gets embarrassed, but we encourage him to get up and shake it off and keep playing. The “Challenger League” is a phenomenal experience for Mason. It is a league that is available for anyone with special needs. It is split up into two groups, 13 years and younger and 13 years and older. There are no real rules, they just let every kid have a chance to bat every inning and everyone have a blast. Kids from the “minor” and “major” leagues will volunteer and help the kids field the ball and run the bases. It is awesome to see the kids’ faces light up after they hit the ball and take off for first base. Mason especially likes to be the last batter because that player gets to clear the bases with an automatic homerun! Mason has recently joined Cub Scouts and has a blast doing it. He’s learning a lot about helping his community. His Tiger pack had a fund raiser and built birthday packages for the local food bank. Through this effort they gathered 362 pounds of food from the community to give birthday cake supplies to those in need! Mason also had a lot of fun with the pine wood derby and took 1st place for the Tiger’s class at the district races.

Having a vision impaired student in the Wyoming schools has been a challenge. Not because they don’t want to help, but there are few vision impaired people in our rural state. There are not many providers and few are familiar with the specific needs of vision impaired students. Mason’s first IEP with the preschool was the most challenging. The short story is they were not providing the vision services that he needed, and we agreed to in the IEP, so we ultimately had to file a complaint with the state. It was a tough process to go through and we spent hours researching special needs education to ensure that we knew what we were talking about as we fought for Mason’s rights. We can’t stress enough that you must be the biggest advocate for your child, no one else will advocate as hard as you will. You need to educate yourself to understand the special education laws and your children’s rights so that they can have the best education that they can get.

Mason is going into the 2nd grade this year and the elementary school experience has been better than the preschool experience. We stole an idea from Mason’s TVI that also has a son with vision loss. We created a flier to help introduce Mason to the school staff and understand his situation. We included pictures, his favorite things, and critical parts of his special needs in simple terms. This was a huge hit, and all of the staff truly cares about him and looks out for him. They’ve made sure that the kids don’t leave things out in the hallway and even added high contrast material to the steps and many other simple things to make sure he’s safe at school. In Kindergarten Mason learned all the letters of the alphabet in print and in Braille, which completely blew us away. He’s also doing better with his orientation and mobility / cane skills. On October 15th, White Cane Day, Mason’s O&M set up activities for his kindergarten class and the other grades to let the other children understand what it’s like to utilize a white cane.

Overall our life has taken us down a path we never thought we would go on, and we wouldn’t change it for anything. We’ve met fantastic people and lifelong friends through the OAA conferences, through our trips to the Children’s Hospital, and through the schools and service providers. If we were to leave you with our biggest learnings on this wild ride known as life it would be:

• Leverage other people that are going through the same things that you are via organizations like the OAA. You are not alone and people going through similar situations want to help.

• Understand the laws and your rights with respect to special education. No one will be a bigger advocate for your child than you. Education is the best tool you can have.

• The greatest quote that we’ve ever heard is “Life is a journey, not a destination” by Ralph Waldo Emerson. You have to make sure that you take time to enjoy those little precious moments in life where your child is in a pure state of joy or does something you never thought they would be able to, like riding a bike!

Brandon and Shandra
Green River, Wyoming
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Allison is 14 years old and Marcel is 18 months old, both were diagnosed with Propionic Acidemia through newborn screening. Allison was hospitalized for a few days right after her diagnosis was confirmed for observation and then again a few weeks later because the doctors were a little too optimistic with her protein tolerance. Both are doing really well! They also have a sister, Mina, who is 3 years old and unaffected and 2 dogs, Freckles and Koda.

Allison likes to play softball and soccer. She likes building with Lego and reading. She loves to draw. Allison plays saxophone in the school band and loves science classes. She is planning on going to college to become a veterinary technician. She loves her siblings and likes to play on the swings or trampoline with them. Allison likes to cook PA friendly meals for the family, mostly veggie tacos, and invite her grandparents over to share it with them. They are delicious! She measures and calculates the protein amounts in her food and makes and drinks her formula by herself every day. Last school year she expressed an interest in going to Washington DC on a school sponsored trip, she raised about half of the money she needed through fundraisers, so we decided to cover the other half so she could go. She had a great time and was very responsible with her diet. We enjoyed seeing all of her pictures and hearing stories from her trip.

Marcel likes to play and hangout with his sisters. His development is on track and he learns more and more every day. His favorite toys are cars and balls. He is learning how to climb everything from his sister.

We are in the process of changing doctors; their long-time doctor has retired so we are changing hospitals, doctors, and dieticians to St. Louis Children’s Hospital. I’m hoping we don’t have to change too much of our process. We have been blessed with the doctors we had and the health of our kids!

Cassie
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HCU/OAA/PAF Conference 2022

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Summer/Fall 2022
Hi my name is Taryn and on January 30, 2000, I was born at 3:15pm at Jefferson Hospital in Philadelphia, Pennsylvania and weighed 7 pounds 1 ounce. When I was born, nothing seemed out of the ordinary. After being brought home, I became very lethargic and couldn't remain awake while breastfeeding. My parents went back and forth with the pediatrician trialing me on different formulas. I vomited with every formula that I tried, and as a result, I started losing weight. In the first week of life, I dropped down to 5 pounds! My mother was at the doctor's office every day and kept telling them that something is wrong. When I was around 8 days old, I was admitted to a local hospital in southern NJ and they had no insight as to what was wrong with me. They thought that I was dehydrated, and that my kidneys weren't working properly. The local hospital called in a nephrologist from Children's Hospital of Philadelphia (CHOP) and after examination, the nephrologist believed that I could have a metabolic disorder, so I was then transferred to CHOP. My mom vividly remembers calling the doctor after she was told this information, asking questions like what is a metabolic disorder? And how serious is this? In response the doctors said that some metabolic disorders can be managed, while others are so severe that the child won't survive. Around midnight on my tenth day of life, I was transferred to CHOP in an incubator riding in an ambulance. My father remembers driving behind the ambulance till this day, crying and fearful, wondering if I would make it or not. Around 1am, we made it to CHOP and the doctors drew blood from my head, arms, feet, legs, and hands. My veins were so small from the dehydration after not eating and vomiting all the time. The next morning, I was diagnosed with Methylmalonic Acidemia (MMA) by Dr. Charles Venditti. To determine the type, I had a skin biopsy sent to McGill University, which later revealed that I had Cobalamin A type.

Following my diagnosis, I needed to follow a low protein diet, and receive hydroxocobalamin injections (1 mg) every other day. As a baby, I used three different formulas: Similac with iron, Propimex, and Pro-Phree. I continued to drink Pro-Phree as supplemental calories until the age of 10. I discontinued the formula when Dr. Venditti informed me that I didn't need the extra calories as I was getting enough calories from regular food. As a young child, my parents told me that getting me to eat was a struggle, and the one food they could get me to eat was French fries. I mean, who doesn't like French fries? As I got older, I started branching out, eating different foods, and wanting to eat more. Then became the struggle of wanting to eat more but wasn't able to due to the low protein diet.

Overall, I had a normal childhood, I tried many different sports, including soccer, softball, ice hockey, and volleyball, but the activities that I have continued with are dance and kung fu. I have always loved dance, especially in the styles of tap and hip hop. I started off doing dance for fun and performing in recitals at the end of the dance season, but when I was in high school, my dance studio closed out, and then I joined a studio that competed, where I won multiple awards with my tap solos, as well as group dances with my fellow teammates. I was able to continue dancing on the dance team as an NCAA Division I athlete throughout my college career, performing at football and basketball games. In kung fu, I have been able to reach 3rd degree black belt and am still continuing my training. I was honored to go on a trip to China in 2018 and train with the Shaolin monks and perform in front of the Abbott of the Shaolin Temple.

After graduating high school in 2018, I attended Sacred Heart University in the Davis and Henley College of Nursing in Fairfield, CT. This was the first time I was living on my own and was a big step, especially in managing my metabolic disorder. Although I had been counting my grams of protein and doing my daily injections while at home, it was a very different experience while in college. While living on campus my freshman year, I had limited options when it came to meals because I wasn’t allowed to have my car and didn’t have the capability to continue next page]
prepare my own meals because all I had was a microwave. Most of the time, I would eat Kraft mac and cheese, ramen, or get a grilled cheese or an egg and cheese breakfast sandwich from the dining hall. Senior year was a little different, as I lived in a hotel for about half the year. I still didn’t have a kitchen but was able to have access to different food places because I was able to have my car. After living in the hotel, I moved into my sophomore dorm room and lived in it for about a month and a half, then COVID hit. School moved to online and remained that way until the end of my sophomore year. To some college kids, moving back home was a struggle, but I saw it as an opportunity. I was able to finish out the year strong because I didn’t have all of those distractions of college, like parties. Speaking of parties, drinking was a new experience where I learned to deal with peer pressure. I felt like I was able to say no in social drinking situations and was able to become the “mom” of my friend group. I always made sure that everyone was being safe and were not getting taken advantage of. Going into my junior year was the start of my clinical hours for nursing school. In my junior and senior year combined, I completed over 500 hours of clinical experience, ranging from 6 hour shifts to 12-hour shifts. During the 12-hour shifts, I had difficulty trying to eat every 4 to 6 hours.

Throughout my 22 years of having this metabolic disorder, doctors tried different routes for my hydroxocobalamin, including varying dosages and frequency. I was initially receiving 1 mg of hydroxocobalamin, then 5 mg, now 10 mg. I also tried taking my hydroxocobalamin orally in formula, but found it was not absorbed as well, so back to injections again. In order to manage my MMA now, my dietary restrictions include 50 to 60 grams of protein per day, as well as daily hydroxocobalamin injections (10 mg) and oral levocarnitine (330 mg twice daily). I also take supplements such as, Vitamin C, calcium, a multivitamin, a probiotic, and DHA. I am currently followed by Dr. Michele Spencer-Manzon at Yale New Haven Hospital Metabolic Clinic.

I live in Ridgefield, CT with my father, Stewart, my mother, Laura, my sister, Nicole, my brother-in-law, Ty, and my brother, Patrick. My boyfriend, who I met my freshman year of college, lives in New Jersey and don’t get to see him very often, but always find time to go onto little trips together. I graduated from college in May, passed my NCLEX (the nursing board exam) at the end of June, and started working as a registered nurse at Stamford Hospital in Stamford, CT in July. Working as a nurse and trying to find time to eat every 4 to 6 hours is difficult so I bring different snacks to eat throughout the day. I live at home and am saving up money in order to get a place closer to work in the future. I continue to train in kung fu and hope to find time to take some tap dance classes.

I attended my first OAA conference in 2010, when I was 10 years old, but didn’t really comprehend everything that was going on. Thankfully, I attended the most recent OAA Conference with my mother, and brother, who just started medical school. I met some amazing people while I was there, like Johnny Tate and his family, where we found out that we were both at CHOP around the same time of diagnoses. I also met Michael Clapcich and his mother who were very welcoming and fun to talk to. In addition, I wanted to thank Dr. Charles Venditti and praise him for all of the research that he has done on MMA and the progress he has made on the development of a cure. I have a greater appreciation and understanding of my disorder, as well as other organic acidemia disorders and hope to continue my learning as a nurse.

My metabolic disorder is part of me, but I have never let it limit me in life.

Taryn Ridgefield, CT taryng77@gmail.com
Destiny
Glutaric Acidemia, Type 1
Age 20

Destiny is a bright and cheerful 20-year-old. She is our middle child of three with an older brother and younger sister who both still antagonize her like any normal sibling (she gives it right back though). But her siblings are also amazing at understanding and helping with Destiny’s needs and knowing sometimes their dad and I must split time between them all when Destiny is at appointments or hospital stays. Destiny has a great sense of humor and a smile that lights up the room. She loves adventure and going on trips. Here is a little bit of her story. Destiny was diagnosed at six months with failure to thrive and during that hospital stay they diagnosed her with GA1. At age 3 she had a cold that caused a metabolic crisis and she had seizures. The seizures left her no longer able to walk, crawl, talk, or eat. But the seizures did not take away her intelligence, her love of adventure, her humor, or her beautiful smile. Destiny has been through many surgeries for her scoliosis, hips, legs, g-tube, and broken bones due to low bone density. Through it all she has kept that beautiful smile (even when she lost all her hair from multiple surgeries in a couple weeks’ time). When Destiny was around 10 years old, we were listening to the radio on the way to one of her doctor appointments. A guest speaker was talking about the Adventures for Wish Kids (now A Kid Again) Christmas Party that would be coming up soon. They talked about their mission of helping make kids feel like a kid again. This was something I knew would be great for Destiny and the rest of our family, so I looked into it as soon as I got home and signed her up. Destiny is aging out of A Kid Again this summer, but it has helped put countless room-brightening smiles on Destiny’s face, as well as smiles on all our faces. Even the faces of foster placements we hosted at the times. A Kid Again have taken us on trips to Magic Mountain, the Columbus Zoo, Kings Island, Charles Penzone, Blue Jackets hockey games, Crew soccer games, Ohio State games, Scene 75, Zoombezi Bay, Holiday Parties and many, many others. They cover tickets to the events, as well as parking and food and usually add in a little surprise too. The Adventures have helped Destiny meet other kids who look just like her (in a wheelchair) and for us to realize we’re not alone. A Kid Again has given us so much joy and happiness and never asked anything in return (though we love to volunteer for them when we are able). A Kid Again has grown considerably since when we first joined and now are throughout Ohio and in many states. We can’t thank them enough. If there is a chance that your child may qualify, please check into www.Akidagain.org. You’ll be glad you did.

Tracy
tjamison16@hotmail.com
MMA produces unique protein modifications that contribute to disease

Dr. Charles Venditti’s lab at the National Institutes of Health has recently discovered a new protein modification in methylmalonic acidemia (MMA) that drives disease pathology. The work of postdoctoral researcher, Dr. Pamela Sara Head, in a recent article published in Science Translational Medicine, demonstrated how the build-up of methylmalonyl-CoA (the substance converted to methylmalonic acid in MMA patients) can also lead to the build-up of a new type of modification of enzymes in liver, kidney, and brain cells. They have termed this modification “methylmalonylation”. Too much methylmalonylation modification on enzymes upsets their normal function often by turning the enzyme off when it should be on or vice versa. Many of the affected enzymes are located in or around the cell mitochondria and when modified can result in MMA symptoms such as hyperammonemia (buildup of ammonia in the blood), hyperglycinemia (buildup of glycine), and poor cellular use of sugar and low energy production. These results were seen in the lab’s mouse models of MMA and more importantly, were confirmed in donated MMA liver tissue from MMA patients who received liver transplants. These discoveries uncovered a mechanism behind the high ammonia and glycine levels that are typically seen in severe MMA and provided deeper insights as to why patients still experience disease symptoms even with around the clock care with protein restricted diets and formulas as well as medical monitoring.

During this study, Dr. Head also discovered which enzyme is responsible for removing the disruptive methylmalonylation modifications. This enzyme is known as SIRT5 and it helps maintain mitochondrial health (keep mitochondria healthy/functioning). After discovering that SIRT5 levels are naturally lower in MMA tissues and determining that SIRT5 itself can also be modified by methylmalonylation, Dr. Head engineered a new version of the SIRT5 protein to escape the abnormal modification and maintain its activity. This “SuperSIRT5” was given to affected MMA mice using a gene therapy called adeno-associated virus (AAV). Treated mice had lower methylmalonylation modifications, increased weight gain, lower blood ammonia levels and restoration of other mitochondrial health markers. Overall, the discoveries by the Venditti lab provide insight into symptom development not just in MMA but potentially in other organic acidemias (OAs) and forms of vitamin B12 deficiency and provide a novel therapeutic target to pursue for future drug development.

For more information you can read the article here: pubmed.ncbi.nlm.nih.gov/35613279/

And the supporting article by Dr. Goetzman and Dr. Vockley here: pubmed.ncbi.nlm.nih.gov/35613282/

We appreciate all the families that have participated in our studies over the years. This study would not have been possible without the families who donated their precious liver samples to our research. More information about our research can be found here: genome.gov/staff/Charles-P-Venditti-MD-PhD
Initial data readout for patients with PA and MMA expected in the first half of 2023.

Interim data from healthy individuals, reported earlier this year, showed that BBP-671 was detected in plasma and cerebrospinal fluid (CSF) at concentrations above predicted therapeutic thresholds, suggesting the compound has the potential to impact key systemic and neurological complications of PA and MMA.

BridgeBio is in active discussions with regulators and expects to launch a pivotal Phase 2/3 clinical study of BBP-671 in pantothenate kinase-associated neurodegeneration (PKAN) in 2023.

If successful, BBP-671 has potential to be a best-in-class therapy for PA, MMA, and PKAN patients, as well as the first approved oral therapy for the treatment of systemic complications caused by CoA deficiencies.

PALO ALTO, Calif., Aug. 18, 2022- BridgeBio Pharma, Inc. (Nasdaq: BBIO) (“BridgeBio” or the “Company”), a commercial-stage biopharmaceutical company focused on genetic diseases and cancers, today announced that the first patient has been dosed in its Phase 1 clinical trial of BBP-671, an investigational oral therapy being developed for the potential treatment of conditions caused by coenzyme A (CoA) deficiencies.

BBP-671 is an investigational oral therapy intended to increase CoA levels by allosterically modulating pantothenate kinases, key enzymes in the CoA biosynthesis pathway. It is being developed as a potential therapy for diseases in which CoA metabolism is deficient, including propionic acidemia (PA), methylmalonic acidemia (MMA), and pantothenate kinase-associated neurodegeneration (PKAN). PA, MMA, and PKAN affect an estimated 7,000 patients in the United States and European Union collectively, with PA and MMA typically diagnosed in early infancy. BBP-671 is based on breakthrough scientific developments from St. Jude Children’s Research Hospital in Memphis, Tennessee.

The first-in-human Phase 1 study of BBP-671 is a single- and multiple-ascending dose study designed to evaluate the safety, tolerability, pharmacokinetics (PK), and pharmacodynamics (PD) of BBP-671. The first part of the study evaluated BBP-671 in healthy individuals and the second part of the study is evaluating BBP-671 in PA and MMA patients. Positive interim data from healthy individuals were reported earlier this year.

The first patient dosed in the second part of the trial has PA. In this part of the study, up to eight PA patients and eight MMA patients will receive BBP-671. Safety and tolerability will be assessed, as well as the PK and PD of BBP-671 using validated bioanalytical assays. A wide range of disease-related biomarkers with potential clinical relevance will be monitored during the study and compared to baseline values. The potential use of biomarkers for PA and MMA as surrogate endpoints in clinical trials for metabolic diseases is a subject of active discussion among key opinion leaders in the field.1

“We are eager to advance the trial of BBP-671 in the hope that it will provide a positive improvement for patients, valuable data, and ultimately lead to a meaningful therapy for patients who currently have no approved treatment options,” said Zineb Ammous, M.D., clinical geneticist and Medical Director of The Community Health Clinic in Topeka, Indiana, which specializes in rare genetic conditions.

PA and MMA are rare metabolic disorders caused by mutations in genes that impact the development of enzymes that participate in amino acid metabolism, leading to life-threatening metabolic decompensations, as well as long-term complications involving multiple organ systems, including the heart, pancreas, kidney, liver, and brain. The current standard of care includes dietary restrictions, supplementation, and sometimes liver and/or kidney transplantation but unmet need remains high due to the long-term and life-threatening impact of these diseases.

“Currently, the majority of individuals with PA and MMA are being diagnosed through newborn screening and are on a strict diet and regimen of supplements and medications. PA and MMA patients experience life-threatening metabolic decompensations, as well as many other serious complications as a result of their disease. We are hopeful that by creating a therapy designed to target the disease by modulating fundamental metabolic pathways, we may be able to treat a condition that impacts young people so early in their lives,” said Jerry Vockley, M.D., Ph.D., Chief of Genetic and Genomic Medicine and Director of the Center for Rare Disease Therapy at
Multiple Ascending Dose (MAD)

Beyond dietary management and liver and/or kidney transplant, and MMA and their families currently have no treatment options associated with improved survival. Individuals diagnosed with PA

Hospital of Pittsburgh, Pennsylvania.

[Image 26x91 to 44x106]

[Image 26x408 to 58x440]

beyond dietary management and liver and/or kidney transplant, and MMA and their families currently have no treatment options

individuals diagnosed with PA

Hospital of Pittsburgh, Pennsylvania.

[Image 26x91 to 44x106]

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Hospital of Pittsburgh, Pennsylvania.

[Image 26x91 to 44x106]

[Image 26x408 to 58x440]

beyond dietary management and liver and/or kidney transplant, and MMA and their families currently have no treatment options

individuals diagnosed with PA

Hospital of Pittsburgh, Pennsylvania.
Are you the caregiver of someone with a chronic medical condition requiring support?

JOIN OUR RESEARCH STUDY!

The goal of our study is to understand how caregivers manage their lives when caring for someone with a chronic medical condition over time.

You may be able to participate if:

• You are 18 years or older
• You are the caregiver of a person with a chronic medical condition or you support someone who cares for a person with a chronic medical condition
• You are willing to share your experiences about caregiving

What will the study include?

• Annual online and phone assessments
• Periodic daily diary logs
• An optional blood sample
• A chance to invite other adults closely involved in your life to participate
• Compensation for your time

The study is completely voluntary and you may choose to stop participating at any time.

If you are interested in participating in the study or have any questions, please call us at 301-219-3394 or email us at CaregivingStudy@mail.nih.gov
Two fellows in Dr. Charles Venditti’s lab spent many hours over several months crocheting what they’re calling Pam and Pauline’s Plushies for Patients.

“These are the coolest handmade stuffed animals I have ever seen,” said Venditti, a senior investigator in NHGRI’s Metabolic Medicine Branch.

Dr. Pam Head, an NIGMS postdoctoral research associate (PRAT) fellow, and Pauline Hoffman, an NHGRI postbac IRTA fellow, made 35 unique plushies for children who were stuck indoors at a medical conference in Bethesda over a weekend in June. The conference was for families grappling with MMA (methylmalonic acidemia), PA (propionic acidemia) and cobalamin deficiencies—rare, genetic metabolic disorders.

Head began crocheting in graduate school to pass the time during lectures. She started making stuffed animals earlier this year as gifts for colleagues returning to the office. When NHGRI clinical investigator Dr. Irini Manoli saw them, she asked whether Head would share a couple with patients visiting the Clinical Center. When she did, the patients’ eyes lit up, recalled Manoli. “It was most heartwarming,” she said.

With the summer family conference in mind, Head then teamed up with Hoffman and the two set to work during off-hours—on weekday evenings and weekends—to prepare one for each child attending the meeting.

“I became not just Pauline’s mentor in the lab but also in crocheting,” said Head.

“It was such a special set of gifts,” said Venditti, who has devoted his career to studying organic acidemias. “All the kids got to pick one and even some of the unaffected siblings got plushies as well.”

MMA and PA are life-threatening conditions that cause frequent hospitalizations and long-term complications such as learning and vision problems. While cobalamin (vitamin B12) injections can help milder forms, treatment largely consists of managing symptoms and, for some patients, organ transplants. Venditti’s team focuses on developing genomic therapies for these disorders.

At the conference, the parents appreciated the plushies that brought their children joy and served the dual purpose of keeping their kids occupied so they could focus on the lectures.

“I hope the person who made them knows how happy the kids were to receive them,” said Misty Garcia, whose daughter received a llama plushie.

Head and Hoffman said they hope to make more plushies in the future to donate to other young patients.

By Dana Talesnik
Reprinted with permission from the NIH record
OAA is excited to partner with AllStripes to create a database that can help researchers better understand the natural course of methylmalonic acidemia (MMA) and propionic acidemia (PA). This database will enable families to have access to their medical records in one secure location while also powering multiple potential future MMA and PA studies.

The first study, JUMP (Journey to Understanding MMA and PA), will help HemoShear Therapeutics gain insight into the disease experience to support development of their potential new therapy. Insights generated by families sharing their medical records on the AllStripes platform will be shared directly with the community through OAA.

To move this research forward, we are looking for 60 individuals impacted by MMA or PA who are willing to contribute their de-identified data from medical records. AllStripes de-identifies the information in your records, by removing information like name and address so your medical journey can be combined with data from the rest of your community to help with research efforts.

Should you choose to participate, you’ll receive research updates and access all your medical records in a secure AllStripes account. AllStripes does the work to collect all your medical records on your behalf, at no cost. Their goal is to remove the burden and expense of requesting these medical records on your own.

1. **Sign up**
   Create your secure AllStripes account and review the research consent (takes about 10–20 minutes) at allstripes.com/mma or allstripes.com/pa

2. **AllStripes gets to work**
   They do all the behind-the-scenes work to collect and process your records

3. **Power research**
   Track your contribution to research projects and receive updates on research insights

**To learn more or sign up, visit:**
allstripes.com/mma or allstripes.com/pa
Frequently Asked Questions

How can my medical records advance research?
Your (or your loved one’s) medical records contain clues that could help advance treatment research. They can be a rich source of knowledge about symptoms, progression and impact of your condition on patients and families. After AllStripes de-identifies the information in your records, by removing information like your name, your medical journey can be combined with data from the rest of your community to help with a wide range of research efforts.
You may also be asked to complete surveys within the AllStripes platform to answer specific research questions or provide more insights back to you and your community.

Why do companies like HemoShear need to learn from patients’ medical data?
MMA and PA are so rare that there is no well-established long-term data on the course of these diseases. HemoShear is collecting natural history data on MMA and PA because the company needs insight into the real world experience of many patients to better understand the disease and be able to scientifically demonstrate whether the potential new treatment they are developing is effective in improving outcomes. The de-identified MMA and PA patient data in the AllStripes platform may be used in the future by other companies who are developing new treatments.

Will I have to pay a fee to join AllStripes, participate in the JUMP study and receive access to my health records?
No. AllStripes’ commitment to patients and families is that there is never a cost to join research programs or use the platform.

How does AllStripes keep patients’ data safe and confidential?
Only properly trained and authorized personnel will have access to patient medical information for processing purposes. AllStripes follows strict policies and procedures to ensure that patient information is protected. To view the full AllStripes Privacy Policy, visit allstripes.com/privacy.

Can I contribute records on behalf of my loved one who has passed away?
Yes. Families who have lost children to MMA or PA can participate in this study. Contributing records on behalf of a loved one who has passed away can create a meaningful legacy and provide valuable information to improve the lives of other patients and their families.

Can I join an AllStripes program if I don’t live in the U.S.?
At this time, AllStripes is approved to collect records and conduct research involving patients in the U.S., Canada and the U.K.
An Update from LogicBio

Happy Autumn! On behalf of the team at LogicBio we hope that your families are keeping well. As LogicBio continues to advance its investigational gene therapy candidate for MMA, we wanted to provide an update on our activities. We are continuing to enroll and progress our SUNRISE clinical trial – a Phase 1/2 gene therapy trial which uses an investigational therapy called LB-001. This trial is primarily looking at the safety profile of two different doses of LB-001 in trial participants, and also the effect of the medicine on biochemical markers of MMA and important clinical features. The most recent updates about SUNRISE are available on our website, www.logicbio.com. If you have specific questions or would like to get in touch with us, please feel free to contact us at patients@logicbio.com.
The HERO (Help Reduce Organic Acids) clinical study, sponsored by HemoShear Therapeutics, is now recruiting participants at 10 leading research hospitals across the United States.

“Participating in clinical trials is essential for making progress to advance potential new treatments,” says Kim Chapman, MD, PhD, medical geneticist at Children’s National Medical Center and lead principal investigator for the HERO study. “We are excited to see that the HERO clinical trial is expanding to more institutions across the country with the hope that the study will be more convenient for families to join.”

HERO is actively recruiting children and adults with MMA (mutase deficient) and PA aged 2 and older who have not had a transplant and meet the study criteria. While in the study, participants can continue to take their medications, including carnitine. More information about the HERO study is provided at MMA-PAHero.com or clinicaltrials.gov.

**Study Assessing Potential New Treatment**

HemoShear is developing investigational drug HST5040 as a potential treatment for MMA and PA. HST5040 is a liquid that is taken at home twice daily by mouth or through a gastric or nasogastric feeding tube. The HERO study is designed to assess how HST5040 acts in the body, if it causes side effects and whether it works to reduce harmful toxins in the body and help people with MMA or PA feel better.

Participants in the study will have the opportunity to continue to take the study drug until it is available to the public or the study is ended.

**Find a Site Near You**

HERO is being conducted at many children’s hospitals across the United States - and more sites are being added. Ask your doctor about whether you could be a candidate to participate.

Transportation can be provided to travel to the study sites and stipends are available to cover meals. All study drugs, study visits and assessments will be provided at no cost.

**Learn more at MMA-PAHero.com**

*The safety and effectiveness of HST5040 for the treatment of MMA or PA have not been established.*
INDICATIONS AND USAGE

CYSTADANE (betaine anhydrous for oral solution) is indicated in children and adults for the treatment of homocystinuria to decrease high homocysteine blood levels. Homocystinuria is a rare genetic disorder in which there is an abnormal accumulation of the amino acid homocysteine in the blood and urine. The following are considered to be homocystinuria disorders:

- Cystathionine beta-synthase (CBS) deficiency
- 5,10-methylenetetrahydrofolate reductase (MTHFR) deficiency
- Cobalamin cofactor metabolism (cbl) defect

IMPORTANT SAFETY INFORMATION

- Hypermethioninemia in Patients with CBS Deficiency: CYSTADANE may worsen high methionine blood levels and accumulation of excess fluid in the brain has been reported. If you have been told you have CBS deficiency, your doctor will be monitoring your methionine blood levels to see if changes in your diet and dosage are necessary.
- Most common side effects were nausea and gastrointestinal distress, based on a survey of doctors.
- To report SUSPECTED SIDE EFFECTS, contact Recordati Rare Diseases Inc. at 1-888-575-8344, or FDA at 1-800-FDA-1088 or www.fda.gov/medwatch.

Visit www.CYSTADANE.com for more information
Summary:
Read this information before you start using CYSTADANE and each time you get a refill. There may be new information. This information does not take the place of talking to your doctor about your medical condition or your treatment.

What is CYSTADANE?
CYSTADANE (betaine anhydrous for oral solution) is a prescription drug used in children and adults for the treatment of homocystinuria to decrease high homocysteine blood levels. Homocystinuria is a rare genetic disorder in which there is an abnormal accumulation of the amino acid homocysteine in the blood and urine. The following are homocystinuria disorders:
- Cystathionine beta-synthase (CBS) deficiency
- 5,10-methylenetetrahydrofolate reductase (MTHFR) deficiency
- Cobalamin cofactor metabolism (cbl) defect

What should I know about using CYSTADANE?
- CYSTADANE is usually prescribed along with other therapies. Individual treatment will be based on the cause of homocystinuria.
- In patients with CBS deficiency (classical homocystinuria), CYSTADANE may worsen high methionine blood levels and accumulation of excess fluid in the brain has been reported. If you have been told you have CBS deficiency, your doctor will be monitoring your methionine blood levels to see if changes in your diet and dosage are necessary.

Before you use CYSTADANE, tell your doctor if you are:
- Pregnant or plan to become pregnant. It is not known if CYSTADANE will harm your unborn baby.
- Breastfeeding or plan to breastfeed. It is not known if CYSTADANE passes into breast milk. Talk to your doctor about the best way to feed your baby if you use CYSTADANE.

How should I use CYSTADANE?
You should use CYSTADANE only as directed by your doctor.
- Shake bottle lightly before removing cap.
- Use the scoop provided to measure the number of scoops as prescribed by your doctor. One level scoop equals 1 gram of powdered betaine.
- Mix the powder with 4 to 6 ounces (120 to 180 mL) of water, juice, milk, or formula until completely dissolved, or mix the powder with food.
- Immediately eat or drink the CYSTADANE mixture.
- Always replace the bottle cap tightly after using.

What are the possible side effects of CYSTADANE?
The most common side effects were nausea and gastrointestinal distress, based on a survey of doctors.
These are not all the possible side effects of CYSTADANE. Tell your doctor if you have any side effects that bother you. You may also report side effects to the FDA at 1-800-FDA-1088.

How should I store CYSTADANE?
- Always replace the bottle cap tightly. This will protect CYSTADANE from moisture.
- Store the bottle at room temperature: 15°C to 30°C (59°F to 86°F).
- Keep CYSTADANE and all medicines out of the reach of children.

General information about the safe and effective use of CYSTADANE:
Medicines are sometimes prescribed for purposes other than those listed in a Patient Information leaflet. You can ask your pharmacist or doctor for information about CYSTADANE that is written for health professionals. Do not use CYSTADANE for a condition for which it was not prescribed. Do not give CYSTADANE to other people, even if they have the same symptoms you have. It may harm them.

What are the ingredients in CYSTADANE?
CYSTADANE is a white, granular, water-absorbing powder for oral solution available in bottles containing 180 grams of betaine anhydrous.

Supplied by: Recordati Rare Diseases, Puteaux, France
Licensed to and distributed by: Recordati Rare Diseases Inc., Lebanon, NJ 08833, U.S.A.
For the most recent prescribing information, please visit www.recordatirarediseases.com/us.
CYSTADANE Labeling Revised 10/2019

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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 an 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