Happy 2021! Hope is on the Horizon!

We have all survived an entire year of living in this pandemic! I hope you all are doing well and I applaud you all for keeping safe and healthy during this time. I’ve heard of many of you whose child contracted COVID 19 over this past year – some managed it better than others. Now, thankfully our children are now receiving the COVID-19 vaccine! I hope all goes well with any possible side effects from the vaccine. My daughter Melissa received the first dose of the Pfizer vaccine last week and did very well. I have included a letter in this issue, written by OAA Medical Advisor, Dr. Kim Chapman regarding the safety of the vaccine.

Rare Disease Day 2021 - this year’s awareness of Rare Disease Day has been a huge success. We would like to thank LogicBio Therapeutics and Hemoshear for their fundraising efforts for OAA. LogicBio offered a matching donation up to $3K – and I’m happy to report we went well above that number! We sold Rare Disease awareness t-shirts (two different styles) – and the donation to OAA was $2,150! Thank you to all that supported this fundraiser (see photos on page 3). Other donations, on Facebook, via our website and checks mailed all contributed well over $5,000! Hemoshear Therapeutics arranged a month long walk/run/bike-a-thon for their employees. Proud of their progress and thankful for their donation to OAA of $5,000!

Finally, I’m sad to report we will need to postpone the conference planned for June this year. Here’s an update from Erin MacLean, OAA Conference Coordinator:

2021 OAA/HCUA/PAF Conference Postponement

The Organic Acidemia Association, HCU Network America and Propionic Acidemia Foundation have decided to postpone the joint conference that was scheduled for June 26-June 27th in Bethesda, Maryland. Since our pediatric population (infants, toddlers and elementary) will not be fully vaccinated for the Corona virus until Fall 2021, we felt we should postpone until every age demographic was vaccinated to have an in person conference. Therefore, we are rescheduling our joint conference to June 2022 in Bethesda, Maryland. Details regarding the 2022 OAA/HCUA/PAF Conference will be forth coming over this year. If you have any questions, please contact Erin MacLean, charlileerinwill2@msn.com or Kathy Stagni, mstagni@gmail.com.

Stay safe,
Kathy
Rare Disease Day 2021 Photos

Clockwise From Left: Propionic Acidemia Family in Georgia, Family celebrating in memory of their sister/daughter in California, MMA CBL C Family in New Jersey, and this year’s tshirt designs
Marc
MMA, CBL B, Non-B12 Response
Age 36

Hi, I’m Marc from Belgium, I have MMA Cbl B, non vitamin B12 responsive.

I’m 36 years old and for a person with my condition, I am in good shape.

Five years ago, I already had the honor to write an article in the OAA Magazine. Now, I am proud to receive this honor again and tell you a little bit about my life and my family.

The article from five years ago was about a big step in our lives. I had just married my wife, Annemieke, a type 1 diabetic. In the article, I told you about how difficult it was for a person with MMA and a person with type 1 Diabetes to commit to the lifelong commitment that is marriage. Two people with a disease who want to live a normal life, two people who took the risk and took this big step in a human’s life. We also announced another big step in our life, because we were pregnant with our first child.

Now five years later, we have taken this giant step. On July 7th, 2016 we had a son, who we called Bengt. “Bengt” translated in Norwegian means “Brave as a bear”. It was a dream come true, this little perfect human being, who was completely healthy came out of 2 people with a disease.

It was a very difficult birth, in which both mother and child had to fight for their lives. But in the end, Bengt honored his name and was brave as a bear and came out as a beautiful young boy. Annemieke also survived the life threatening experience.

Now, we have a little miracle, a healthy and energetic son. Yes, it was a risk to have a child, not knowing what our own future will bring. How our disease will evolve, would we be able to take care of Bengt? Would we be able to give him a future? But we are so happy we took the risk!

Bengt is an active, sweet, little boy who gives us more joy in life than we have ever had before. Although he can make us tired, even completely exhausted, his smile and enthusiasm also gives us more energy than any energy drink would ever give us.

The combination family – work – health really became too much for me. I moved to working parttime. My kidneys became worse. With a lack of patients with MMA in Belgium, we went to the NIH in Bethesda to Dr. Manoli and Dr. Vendetti, in 2019, to ask for their opinion. Finally the result came and my kidneys were effected, but I didn’t need transplant right now, but I will in the near future. My liver was ok. So, it would only be a kidney transplant and not a kidney-liver transplant.

Because we like to travel, after our visit at the NIH we went travelling the USA for a few weeks and we visited family in the USA. After going through China this was the 2nd big trip for Bengt in his short life.

The big question remains what the future will bring? Are we going to be able to give Bengt the future he deserves? Are we going to be there for him when he needs us? Aren’t we going to be too sick or too tired if he needs us, I don’t know. Maybe all parents ask themselves these questions, but we did think this over and we are sure we will do anything in our power to give Bengt anything he needs. And because we’ve been through so much in our lives, we realize even more than other people, that it is the little things in life that matters the most and being near the people who love you is the most important. And that is the message in life we want to give Bengt.

Our diseases could be an enrichment in his education, instead of an obstruction. We raise Bengt with the message to enjoy every day and savor the big moments in life, but also enjoy the many little moments of joy in his life.

Don’t get us wrong, having a child while you both have a disease is a challenge. But with the support of family and friends it may work for us. Bengt has two parents who will do everything to make him happy, now and in the future.

Maybe the message we want to give you is that sometimes it is worth taking a risk!

Marc, Belgium
marc_for_fcb@hotmail.be
Isovaleric aciduria (IVA) is an organic aciduria presenting with a severe (classic IVA) or attenuated phenotype (mild IVA). For more than 20 years, IVA has been increasingly included in newborn screening (NBS) programs worldwide with the implementation of tandem mass spectrometry. Since the long-term clinical benefit of screened individuals was only rarely investigated, we conducted a national, prospective, observational, multicentre study of individuals with confirmed IVA identified by NBS between 1998 and 2018 in Germany (Mütze et al. 2021).

The participating German pediatric metabolic centers reported to follow 128 screened individuals with IVA identified by NBS since 1998. About 80% (N=102) of them are supposed to follow an attenuated disease course (mild IVA), while 20% (N=26) are classified as classic IVA. Ninety-four individuals with IVA, representing 73.4% (for classic IVA: 92.3%) of the German NBS cohort, participated our long-term observational study.

Compared to the pre-screening era ((Grunert et al. 2012) mortality: 18.7%; (Tanaka 1990); mortality: 43.2%), mortality in screened individuals with classic IVA (N=24), the overall clinical outcome was favourable, but NBS did not completely prevent metabolic decompensations (N=17), 76% of them occurred already in the first days of life. IQ (mean ± SD, 90.7 ± 10.1) was mostly normal but below the reference population (P<0.0022) and was even lower in individuals with severe neonatal decompensations (IQ 78.8 ± 7.1) compared to those without crises (IQ 94.7 ± 7.5; P=0.01). Similar results were obtained for school placement.

In contrast, individuals with mild IVA had excellent neurocognitive outcomes (IQ 105.5 ± 15.8; normal school placement) and a benign disease course (no metabolic decompensation, normal hospitalisation rate), which did not appear to be impacted by metabolic maintenance therapy.

Examining the largest and longest continuously followed cohort of IVA patients identified by NBS (Mütze et al. 2021) we could confirm that NBS reduces mortality in the classic IVA group and allowed a favorable neurocognitive outcome, but did not reliably protect against (neonatal) metabolic decompensations. Long-term observation of the large group of individuals with mild IVA identified by NBS showed that they do not certainly benefit from NBS. Since their excellent neurocognitive outcome was not explained by therapy, harmonized stratified diagnostic and therapeutic concepts for classic and mild IVA identified by NBS are urgently needed. Finally, the study highlighted that an accurate assessment of the clinical benefits of NBS programs requires long-term observation and careful consideration of disease variants.


I had a relatively smooth pregnancy and birth, so the whirlwind of Santiago’s first couple weeks of life were quite unexpected.

It was discovered Santiago was breech the day before my due date. I was scheduled for a C-section the next morning on August 20, 2020. My husband Rodrigo and I were living in the small city of Prince Rupert, on the north coast of B.C., Canada in the Great Bear Rainforest, where I was working as a Registered Nurse. I had the strange but somewhat comforting experience of giving birth surrounded by my colleagues at the 25 bed hospital at which I had previously worked. My C-section went smoothly. Santiago was exactly full term, weighed 3.72kg, appeared healthy, and was breastfeeding relatively well.

On the morning of August 22, 2020, the nurse took Santiago to the nursery to give me some much needed sleep. I woke up a few hours later and Santiago had not been returned to my room. The nurse had discovered that he was breathing rapidly, so she had called in the locum pediatrician to assess him. Santiago’s newborn screening had been done 24 hours after his birth but the results were not ready yet. What we did not know at the time was that his rapid breathing was his way of blowing off excess acid from his body in the form of CO2. This was to compensate for the metabolic acidosis he was entering.

I was stunned when I went into the nursery to find Santiago under the warmer with an IV infusion in his arm. The pediatrician told us that Santiago was not doing very well but she was not sure what was wrong. She ordered bloodwork and a chest X-ray for him. She queried that perhaps he had sepsis. It was determined that the best plan would be to medevac Santiago to the NICU in Prince George, northern BC’s largest centre.

The journey out involved many steps and was difficult for me with my fresh cesarean. Santiago and I were brought into an ambulance, which took us to a helicopter, which flew to the airport on a nearby island, on which we boarded a tiny plane that took us to Prince George. The plane was too small for my husband to join, so Rodrigo drove the 8 hours to Prince George. My mom lived in Prince George but had come to stay in Prince Rupert for some months to help us out with Santiago, so she joined Rodrigo on the trip. My mom had not yet been able to meet her grandson because of the hospital pandemic precautions.

Santiago had lots of bloodwork done in Prince George. Sepsis was ruled out and it was thought that he probably had some kind of metabolic disorder. The pediatricians asked us whether there were any incidences of metabolic disorders, SIDS, or infant deaths in our families (there are none), or whether Rodrigo and I are related by blood (we’re not).

Santiago’s bloodwork responded poorly when we tried breastfeeding again, so he had to keep receiving his nutrition through IV infusion only. He had a nasogastric tube inserted as a precaution and in preparation for possible tube feeds. I pumped every 3 hours or so to establish my milk supply. A diagnosis could not be determined from the relatively small hospital of Prince George, it was decided we had to be medevac’d again, this time to B.C. Children’s Hospital in Vancouver.

Rodrigo drove another 9 hours from Prince George to Vancouver after Santiago and I were medevac’d. Santiago and I arrived around midnight to a bright, clinical NICU room at the BC Children’s Hospital. I felt dazzled by the state of the art facility but also disconcerted that Santiago’s level of care had to be elevated.

The Biochemical team introduced themselves to us the next day. Eventually Santiago was able to feed a special mix of medical formulas and expressed breast milk orally every 3 hours, while still keeping the IV running. The dieticians were continuously amending his formula recipe in response to his bloodwork. His frequently-used veins led to many failed “pokes” and caused him a lot of upset.

A couple of days after our arrival, the Biochemical team doctors gave us the diagnosis of isovaleric acidemia (IVA). A part of me was relieved to have an answer as to what was wrong with Santiago. Another part of me was incredulous that this mysterious disorder I had never heard of was affecting my baby.
Even with the diagnosis, we really struggled to understand what life for Santiago would look like. The Biochemical team’s doctors, dieticians, and nurses gave us thorough education, but in our sleep-deprived and stressed-out states we kept having to ask that information be repeated to us again because we had so much trouble comprehending. We had difficulty understanding what his lifelong diet would look like and how a person could be expected to grow “normally” with such a protein-restricted diet.

Santiago was discharged on September 1, 2020, after his bloodwork stabilized and a head ultrasound ruled out any brain damage from having had high ammonia levels. The dieticians determined his suitable formula recipe and he demonstrated he could tolerate the required volume of his feeds. He was put on glycine and levocarnitine for medication and Pro-Phree and I-Valex 1 in his formula. He would be unable to nurse because his protein intake needed to be monitored. I opted to exclusively pump as a way to give him a measured amount of breastmilk.

We had only been hospitalized for about 2 weeks, but it felt like months! Now we had to make our way back to our home in Prince Rupert. The best route was still a challenging one to do with a fresh-from-the-NICU baby--taking a 3 hour ferry from Vancouver to Vancouver Island, spending a night in a hotel, driving 7 hours to another city on the other side of the island, and then boarding a 20 hour ferry. Throughout these various legs of the trip Santiago had to feed every 3 hours and I had to pump every 3 hours. We were terrified he would suddenly decompensate on the ferry, on which there is no cell service and we would be unable to reach the Biochemical team for guidance. Thankfully, Santiago did very well on the trip. In Prince Rupert, my parents were finally able to meet their grandson.

Santiago’s health remained stable after our return home. He had one period of poor feeding which led to an ER visit, where it was discovered his bloodwork was a little off. He improved after a couple days of feeding his sick day formula in smaller volumes every 2 hours.

Santiago needed weekly weights and regular bloodwork. The bloodwork proved quite challenging at the rural hospital that was unaccustomed to pediatric patients and to sending out ammonia samples. His blood draws were usually very difficult and lengthy. On more than one occasion the sample was incorrectly shipped out for analyzing. The results were always high but deemed to likely be “false” because his clinical presentation was stable.

Despite Santiago’s seemingly good health, we were disturbed by the fact we were never able to get accurate ammonia results. We also knew there would inevitably come a time when he would get sick and need specialized care which was unavailable in Prince Rupert. We decided to move to Vancouver to be close to the BC Children’s Hospital. We had lived in Vancouver for many years so we were familiar with the city. We moved in December, 2020 when Santiago was 4 months old. To this day, Santiago has been very stable, but it is a huge relief to know that if something happens, the Biochemical team is just a short drive away!

Santiago did not suffer a severe metabolic crisis, thanks to the newborn screening and the dedicated, competent Biochemical team and other healthcare professionals involved in his care. Our main point of contact in his care has been his dietician, who is incredibly patient and responsive to our needs.

I like to say Santiago is special because he is always going against the norm--only 4% of term babies are breech, only 4% are born on their due date, and only 1 in every 250,000 has IVA. I am also pretty sure most babies don’t ride in an ambulance, helicopter, two medevac planes, and overnight ferry all within their first couple weeks of life! Santiago is resilient, bright-eyed, and playful, and I am sure he will keep surprising us.

Andrea, Vancouver, B.C.
azcona.axen@gmail.com
O
tober 13, 2005... The day my first beautiful daughter Michaela came into the world was one of the most blessed days of my life. I loved to stare at her big brown eyes, full head of black hair and all her little fingers and toes. I couldn’t get enough of staring at her and admiring the creation she was. Two weeks into her life we received that call that no mom wants to hear. We were told that Michaela’s new born screening test had triggered. We were advised that she and I needed to have further testing done immediately to determine which of the metabolic disorders she may have. They started her on levocarnitine immediately. They told us to shelter her and not expose her to germs. They told me to start a low protein diet. All that baby weight fell off in an instant and I began losing more weight beyond what I should have ending up around 100lbs. I remember holding Michaela when I found out and praying to the Lord that He would let us switch places. A strange prayer now in retrospect. When I was younger I thought I should bargain with God. Silly me. I should have been praying health for BOTH OF US. Two weeks later the phone rang and I heard the only words I wanted to hear..."Michaela is not positive for any metabolic disorders. She is just a carrier.” The doctor went on to say, “however you have tested positive for 3MCC.” In that moment I didn’t care. I fell to the floor and cried. I was alive at 30 years old and had not made any connections that my health issues through life in the moment could have been related to this 3MCC. I was advised to see a specialist at Boston Children’s which I did. Because my carnitine was extremely low they put me on what was supposed to be a “therapeutic dose” of levocarnitine. This made me extremely sick in combination with a low protein diet. I was not nutritionally supported the way I needed and the carnitine was wreaking havoc with my system at that dose.

In retrospect I had health problems that doctors could not explain. I was weak as a child. I didn’t walk until I was about 18 months. I would fatigue easily. I had a lot of allergies and nearly bled to death when I started my period at age 14. I had nose bleeds that were uncontrolled as a child and struggled with bouts of anemia. As an adult I had every test possible. At one point I ended up in the hospital unable to absorb foods well, weak and having tremors. I remember the doctors standing around me and basically saying to me...this is all in your head like this was a mental illness. I refused all carnitine while there. I cleansed my body and after about a year of supplements and real food again I started to recover fully. I left the carnitine and treatment options in the dust because I could not believe how sick I had gotten. I became untrusting of this medical system that was supposed to help me. Many years later when I was about 37 all the symptoms began again and I decided this time to find a new team. I went to Dartmouth in NH. I found an amazing team there who helped me for the first time truly understand 3MCC, how it worked and what treatments now would help. We started the levocarnitine at a much lower dose than it had been initially prescribed and that was more than enough to get me into the right blood level range without harming my system. I went on 660mg vs 1980mg per day. I was started on X-Leu Maxamum which is a protein supplement from Nutricia America that allows me to get ample amount of protein without the leucine. This time around...
12 years later, Nutricia America had insurance specialists to navigate the complexities of the medical world and coverage. I was a medical social worker and for years I could not get this product covered through insurance on my own. I would sit there and sob not just for me but more so for moms who only knew they had a sick child and had no idea how to navigate a medical insurance system with pre-authorizations, scripts and supporting clinical notes. I vowed then to make sure if I found the answer, no other mother would struggle the way I did trying to help themselves or their babies. With this formula and the levocarnitine, I have been able for the first time in my life to eat enough protein to actually be able to build muscle mass! I can exercise and walk miles. I can hike, bike and climb mountains. All of these things before would put me down for days as my body tried to recover. I am so thankful how far science has come with all of these metabolic disorders since I was diagnosed. At that time in 2005, the doctors were telling me from what they could tell I was the 13th person in the world with it. I thought boy…lucky me. Now we know it is far more common. There are lots of people among us walking around asymptomatic. For those of you who were diagnosed even as adults and have had lifelong health problems- I have no doubt that there will be many more adults showing up with lots of wisdom to share. This will help us all as we age and more and more people are finally diagnosed. It will be so helpful to know each other’s stories and share resources. I vowed to myself that no mom, dad, family member or human would have to go through this journey alone and scared like I did for many years. This is my way of giving voice to all of you who may be reading this for the first time and wondering “where do I begin?” You are not alone. WE are many. Together we are strong.

Heidi, Jericho, Vermont
heidiabair@gmail.com

Medical Insurance Support

Compassion Works Medical – CWM, has been around for years advocating, teaching, and supporting all clinics and patients with the challenges of medical food coverage for people with rare genetic diseases, plus much more. CWM would be your personal reimbursement hub. We would help alleviate all of your medical food coverage frustrations, so you could focus more on your health. In addition to educating you solely on Medical Food insurance terminology, CWM is a non-profit organization and we do not charge for our services. Supported donations are appreciated.

Please feel free to visit CWM at compassionworksmrs.com for more information. For your convenience, we have added a medical food insurance coverage presentation to help educate others.

CWM has expanded their services and are now partners with Hearts Enteral a dedicated DME supplier. CWM and Hearts Enteral helped create a system to help patients with rare genetic diseases afford and stay on diet by providing an all-in-one place you can trust. Please visit Hearts Enteral at heartsenteral.com to learn more.
Hello,

As COVID-19 continues to impact all of our lives, there is some hope on the horizon with the introduction of several extremely effective vaccines from multiple sources. The speed in which these vaccines have been designed, tested and produced is truly amazing and due to the diligent work by many dedicated people. We will look back at this momentous effort as a true miracle of modern science.

Our next task, as a population is to encourage and participate in the widespread vaccination of everyone in our population so that new COVID-19 infections will drop and we will be able to return to interacting with others in person outside our household. Only widespread vaccination, estimates are at least 50%, but more likely 70-90% of the population needs to be vaccinated, will protect us all.

According to science’s current knowledge, having an organic acidemia (of any type) does not preclude getting the COVID-19 vaccine and so we encourage everyone to get the vaccine when they are able. There will be some individuals who cannot get the vaccine (discuss with your own doctor if you have concerns), however having an organic acidemia is not of the reasons.

For those who have a protein restriction, neither of the COVID-19 vaccines being produced by Pfizer or Moderna contain any protein.

The Moderna and Pfizer COVID-19 vaccines are a very new kind of vaccine (for infectious diseases—the technology have been used to help treat cancer in the past). These vaccines are made of RNA (ribonucleic acid) protected by a coat of lipid (naturally occurring fats). RNA is the building block our own cells use as a code for producing important proteins. The cells read RNA code and build new proteins using amino acids already in your body. After the vaccine is injected into the muscle, the muscle cells take up the RNA-lipid particles and strip the lipid coating off. The RNA code then teaches the muscle cells to produce a single protein from the COVID-19 virus. The code provided is the code for the COVID-19 spike protein, one protein this is found on the outside of the COVID-19 virus particle. The muscle cells will transiently produce the COVID-19 spike protein (but not the whole virus) for just a few days until the RNA code spontaneously degrades. The spike protein is ‘presented’ to the outside of the muscle cell wall where the immune cells circulating in the blood can see it. Since it is a foreign protein not normally found in our bodies, the immune cells recognize the spike protein as foreign and start gathering everything needed to destroy the protein. A second injection of the vaccine three weeks later really solidifies the immune response and commits it to the memory of the immune system. Thereafter, if the individual encounters the actual COVID-19 virus which is covered in spike proteins, her immune system will immediately recognize the foreign spike proteins and reactivate to destroy the virus. I have reviewed the data provided by Pfizer and Moderna to the FDA on the efficacy of their vaccine; it is remarkably effective.

Traditional vaccines, like the influenza vaccine, are based upon injecting the actual virus which has either been altered to not cause severe disease or has been killed. The vaccine still carries the viral proteins that the immune system will recognize as foreign. Traditional vaccines do contain protein, but the amount of protein injected is very small compared to the amount of protein consumed even in the most strict organic acidemia diet. Even though traditional vaccines contain protein, we still advise everyone with an organic acidemia to receive all recommended vaccinations.

To conclude, the Pfizer and Moderna COVID-19 vaccines do not contain any protein. They contain a code that will allow the transient production of a very small amount of a virus protein from amino acids already in the body. They will not affect metabolism. It is crucial that all of us participate in the COVID-19 vaccination program in order to defeat this pandemic. I encourage everyone with an organic acidemia to participate.

The CDC and your state’s government’s Board of Health have the most up-to-date information for vaccination plans in your area. Please be patient, much of the information about the tiering system that prioritizes vaccine delivery is on your state’s government’s Board of Health website, but I can confirm that this can change daily depending on the availability of vaccine in your area.

If you have questions regarding the vaccine for you reach out to your Metabolist and potentially primary care physician.

Thanks so much.

Kimberly A Chapman, MD PhD
Geneticist and Metabolist
Medical Advisor Organic Acidemia Association

December 28, 2020
Do you find yourself in the hospital frequently?

WE CAN HELP!

The Reeder family is grateful for the Organic Acidemia Association and the continued support and knowledge they gain from being part of such a wonderful community. Their 6 year old daughter, Myka Joy, is diagnosed with Methylmalonic Acidemia and had a liver transplant in 2017. Being no strangers to hospital life and ongoing medical issues, the Reeders founded Pressing On in 2019, a non-profit aimed to support families experiencing crisis by providing resources and support. Pressing On is based out of Pittsburgh, PA but provides support for families all across the United States.

Check out our website for more FREE Resources & Programs for your family:

www.PressingOn.org

WE CAN HELP!

Our family has been through some tough times over the years, including the journey of a life-saving liver transplant. We know that long-term hospitalizations and critical illnesses are difficult, so we started a non-profit organization to come alongside families in crisis. It’d be our privilege to connect with you during your tough time!

www.PressingOn.org

HOSPITAL PACKING CHECKLISTS

- Planned Hospitalization
- Emergency Room Trip
- Ronald McDonald House Stay

Take the guesswork out of packing! Download any of our free, custom checklists to pack and prepare for your time at the hospital.

Tunes for Tough Times

Stream on Spotify & YouTube

MUSIC PLAYLISTS FROM PRESSING ON

www.PressingOn.org
Dear Parent/Patient:

Starting in September 2020, we’re introducing some exciting improvements to our current Abbott Level-2 metabolic medical food products.

These improved products will still meet your child’s or your unique nutrition needs, but now they will include new ingredients to further support growth and development.

**HERE’S WHAT’S IMPROVING:**

- **NOW HAS DHA**
  to support brain and eye health

- **INCREASED VITAMIN D**
  to support strong bones

- **NON-GMO**
  to be the only metabolic medical foods that are non-GMO

These updated products will still provide the same quality nutrition you’ve come to expect from Abbott. The mixing instructions will not change, and the products will still be halal and made with kosher ingredients.

Due to these improvements to the formulas, you or your child may notice a difference in smell, taste, or color. We are here to help and are partnering with your health care team to provide a transition guide to these improved products if needed.

**FOR QUESTIONS REGARDING THE AVAILABILITY OF THESE UPDATED PRODUCTS, PLEASE CONTACT YOUR SUPPLIER.**

For more information about the formula updates, please contact your health care provider or Abbott Consumer Relations at **800-986-8505**.

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* 900-1100 IU vitamin D/100 g powder compared to previous Abbott Level-2 metabolic formulation with 300-325 IU vitamin D/100 g powder.
† Ingredients not genetically engineered.
‡ Not declared on competitor product websites as of June 30, 2020.
The past year has been consumed by the pandemic. But it was also one that did have some great achievements especially for our rare inborn errors of metabolism. This past December, I had the distinguished honor of receiving the Rare Voice Award for State Advocacy for Rare Diseases by the Every Life Foundation. The award is a beautiful “Abbey” statuette commissioned specially for the Rare Voice Awards, and named for Abbey Meyers, founder of the National Organization for Rare Disorders (NORD). The organization’s mission is to empower the rare disease community to advocate for impactful, science-driven legislation and policy that advances the equitable development of and access to lifesaving diagnoses, treatments and cures.

I have been advocating for expanded newborn screening and rare diseases for eighteen years, after my son Stephen received a late diagnosis of isovaleric Acidemia (IVA) at age 3 ½ due to lack of newborn screening for IVA. The late diagnosis resulted in a traumatic brain. A year later, my daughter Caroline was born and she was diagnosed early with IVA sparing her the same outcome.

This significance of this award is that I received this award for my work with Del. Kathleen Murphy, a local delegate and champion for rare diseases here in Virginia. Our goal was to pass a bill that would mandate insurance coverage for medical formula and foods for these rare inborn errors of metabolism and we succeeded! This is a major milestone here in Virginia after several attempts to pass such legislation over the past fifteen years failed. Prior to this bill, families with a family member needing these lifesaving forms of treatment for their disorder faced tremendous financial hardship if their insurance companies refused coverage for them. Many insurance companies either provide partial coverage or no coverage at all due to the rarity and cost despite the medical necessity of these formulas and foods. This has been a problem around the country as Federal Policy has failed to pass such legislation leaving it up to each state to develop their own policies. Del. Murphy also received the Rare Voice Award for State Legislator for her work with the bill.

I am grateful and humbled to receive this recognition and award for my advocacy work, but more importantly for what it represents for our rare inborn error of metabolism community. I am proud to be able to represent OAA in all of my continued advocacy work.

“I am grateful and humbled to receive this recognition and award for my advocacy work, but more importantly for what it represents for our rare inborn error of metabolism community. I am proud to be able to represent OAA in all of my continued advocacy work.”
LogicBio Therapeutics is pleased to provide an update to the Organic Acidemia Association on our activities over the past several months. Most importantly, we hope that OAA members and their families are keeping well during these unprecedented times. It’s hard to believe that it has been a full year since the Coronavirus pandemic reached the United States. Despite the impact of COVID-19 and the many challenges we have all faced over the past year, the team at LogicBio has remained focused on our research program to develop a new treatment for methylmalonic acidemia and building educational resources for the MMA patient community.

First, we would like to provide an update on our Phase 1/2 SUNRISE clinical trial for MMA caused by a mutation in the MUT gene. This clinical trial will assess the safety profile of two different doses of LogicBio’s investigational gene therapy, hLB-001, in trial participants ages 6 months to 12 years as well as the effect of the medicine on biochemical markers and clinical features of MMA. At the time of the writing of this article, we are excited to announce 4 trial sites are now open and recruiting study participants (Children’s Hospital Colorado, Seattle Children’s Hospital, UPMC Children’s Hospital of Pittsburgh, and Vanderbilt Children’s Hospital) with several additional sites expected to begin recruiting in the next few months. If you would like to learn more, please visit www.sunrisemma.com where you can find more detailed information including educational videos and Frequently Asked Questions. We also encourage you to speak with your child’s physician.

In early January, we had an opportunity to offer a virtual educational session in partnership with the OAA. This event included an overview of gene therapy and an update on LogicBio’s MMA program presented by Dr. Daniel Gruskin, LogicBio’s Head of Clinical Development followed by a Q&A session with attendees. The webinar recording is available via the OAA website. During the program we also introduced our new Understanding Gene Therapy educational video which is available for viewing on the LogicBio website at logicbio.com/patients-families. We are very grateful to Kathy Stagni and the OAA for hosting us and to everyone who joined for this update and discussion.

In addition, despite the virtual nature of this year’s Rare Disease Day events, the team at LogicBio found several ways to recognize the day and pay tribute to the millions of families living with rare diseases globally. We were honored to host Melanie and Ayla Wiseman for a virtual event with the LogicBio team to share their experience with MMA. Melanie and Ayla helped our employees to have a deeper appreciation for what it means to live with MMA and graciously answered many questions from the group. From the practical day to day management of MMA to the physical and psychosocial effects, their talk will not soon be forgotten. We thank them for...
We are also happy to announce the OAA matching gift campaign’s successful close. LogicBio was pleased to provide OAA with the full $3,000 donation in recognition of Rare Disease Day and in support of the OAA’s critical work in the community. The LogicBio team also participated in the #Showyourstripes campaign sponsored by the National Organization for Rare Disorders. This campaign was a major event on social media for much of February 28th, helping raise awareness of rare diseases all around the world.

At every stage of our work, the team at LogicBio recognizes the importance of our connections to the MMA community and the perspective that families bring to us about their experiences. If you are interested in learning about opportunities to connect with us including participation in our parent advisory board meetings or other ways to share your feedback with us, please contact our patient advocacy group at patients@logicbio.com. Thank you.

LogicBio Therapeutics is a biotechnology company committed to delivering genetic medicine to pediatric patients with rare diseases and their lead development program is in methylmalonic acidemia.

They are also committed to understanding the family and caregiver experiences of caring for a child with MMA; they know that the more they learn from the community, the better they can represent the patient and family perspective and focus on developing the right resources and programs. In this spirit of community collaboration, LogicBio is planning for a one-time caregiver focus group in early June. The goal of this virtual event is to hear directly from families, seek feedback on educational resources and learn more about community partnering activities.

Specifically, they are looking to hear from:

- USA-based parents of children under the age of 15 who have been diagnosed with Methylmalonic Acidemia caused by MMUT mutations
- A mix of families who have gone through a transplant and those who have not.
- One parent/caregiver per family.
- Individuals will be paid a modest stipend for their participation. If this is an opportunity you or someone in your family may be interested in, please email patients@logicbio.com to learn more.
OAA/PAF INVITE YOU TO JOIN

Advancing mRNA to Treat Organic Acidemias
Guest Speakers From moderna

📅 Saturday, April 10th | 11am EST

MATTHEW LUMLEY, MD, PhD
Sr Director, Rare Disease Clinical Development

STEPHEN GLEASON
Director, Clinical Operations

OAA/PAF host an informational webinar for MMA/PA Families to learn more about Moderna’s potential treatment and clinical trials

MODERNA Register at: https://us02web.zoom.us/webinar/register/WN_elLs2TA1S6eXCFt-F7lpwg

New Horizons for MMA and PA
Current Treatments, Clinical Trials and New Approaches

A Webinar for Caregivers
Saturday, April 17 1pm - 2:30 pm Eastern
(10 am - 11:30 am Pacific)

HEMOSHEAR THERAPEUTICS

HEMOSHEAR Register at: https://us02web.zoom.us/webinar/register/WN_23hrLYLoR_6o8DlfGaNldA
Recordati, through its US subsidiary Recordati Rare Diseases Inc., today announced the U.S. Food and Drug Administration (FDA) has approved a new indication for CARBAGLU® (carglumic acid) tablets 200mg as an adjunctive therapy to standard of care for the treatment of acute hyperammonemia due to propionic acidemia (PA) or methylmalonic acidemia (MMA) in pediatric and adult patients.

CARBAGLU is the first and only FDA approved medication for the treatment of acute hyperammonemia due to PA and MMA. CARBAGLU was initially approved by the FDA for N-acetylglutamate synthase (NAGS) deficiency, another rare metabolic disorder, as adjunctive therapy to standard of care for the treatment of acute hyperammonemia due to NAGS deficiency, and maintenance therapy for the treatment of chronic hyperammonemia due to NAGS deficiency.

PA and MMA are rare inherited metabolic disorders that result in the dysfunction of a specific step of amino acid catabolism, or breaking down of certain fatty acids, due to deficient enzyme activity. As a result, toxic metabolites accumulate, which can cause hyperammonemia, a potentially life-threatening condition. Carbaglu acts as a replacement for N-acetylglutamate (NAG) in NAGS deficiency, PA, and MMA patients by activating carbamoyl phosphate synthetase (CPS 1), improves or restores the function of the urea cycle, and facilitates ammonia detoxification and urea production.

“There are few approved drugs that treat hyperammonemia, and none that are indicated for the treatment of acute hyperammonemia in PA and MMA patients”, said Mendel Tuchman, MD, Medical Geneticist and Professor Emeritus of Pediatrics at The George Washington University School of Medicine and Health Science. “Carbaglu has the potential to impact these patients by reducing high plasma ammonia levels during critical situations.”

FDA approval of the new indication was supported by a randomized, double-blind, placebo-controlled, multicenter clinical trial comparing the effectiveness of CARBAGLU to placebo in the treatment of hyperammonemic episodes in patients with PA or MMA. The efficacy evaluation, based on 90 hyperammonemic episodes occurring in 24 patients, showed that patients receiving CARBAGLU demonstrated a quicker reduction of ammonia compared to patients receiving placebo. The primary endpoint was the time from the first dose to earlier blood ammonia level below 50 micromol/L or hospital discharge. Throughout the first three days of treatment, a higher proportion of CARBAGLU-treated episodes reached the primary endpoint compared to placebo-treated episodes.

In the clinical trial, at least 1 adverse reaction was reported in 42.2% of the 90 hyperammonemic episodes that occurred. The most common adverse events were (≥5%) neutropenia, anemia, vomiting, electrolyte imbalance, decreased appetite, hypoglycemia, lethargy/stupor, encephalopathy and pancreatitis/lipase increased.

“People living with these conditions face potentially serious complications from hyperammonemia that, without treatment, can lead to coma or even death,” said Andrea Recordati, CEO. “We are pleased to receive this new indication for Carbaglu which will enable Recordati to address an unmet medical need in these patients.”

About Propionic Acidemia (PA) and Methylmalonic Acidemia (MMA)

Propionic acidemia (PA) and methylmalonic acidemia (MMA) are rare inherited metabolic disorders. PA results from a deficiency of the enzyme propionyl-CoA carboxylase. Most PA patients present with symptoms in the first few days of life.

MMA results from different types of enzyme deficiencies or defects. The most common cause is a deficiency of the enzyme methylmalonyl-CoA mutase. In both disorders, complete lack of enzyme or very limited enzyme causes more severe symptoms.

Hyperammonemia is one of the most severe and life-threatening events that can occur in patients with PA or MMA. Hyperammonemia is a medical emergency that if left untreated, can progress to irreversible brain damage, coma, or death. For more information, please visit www.recordati.com.
People who are diagnosed with PA are missing the propionyl-CoA carboxylase (PCC) enzyme, which is responsible for the breakdown of certain proteins and fat. When this enzyme is missing, it is difficult for cells in the body to turn food into energy, leading to toxins building up in the bloodstream. Currently, PA is treated by lowering the amount of protein eaten daily, taking dietary supplements or antibiotics, or getting a liver transplant.

The Paramount Study is a Phase 1/2 study. It is designed to evaluate if an investigational treatment called mRNA-3927 is safe and effective in reducing the symptoms of PA in individuals one year of age and older. mRNA 3927 is an investigational intravenous (IV) infusion treatment that instructs a person’s body to make a PCC enzyme that works.

More information including full trial inclusion and exclusion criteria can be found at trials.modernatx.com or by visiting clinicaltrials.gov.

“As a company, we often talk about the societal responsibility we feel to deliver on the promise of mRNA science for patients. That sense of responsibility is felt with great intensity, and urgency, by the Rare Diseases team. We understand that, for so many patients suffering with a debilitating rare disease, there are no approved treatments. We are committed to leveraging our mRNA platform to advance medicines for some of these diseases and bring new hope to patients and their families.”

- Paolo Martini, Chief Scientific Officer, Rare Disease
Dear MMA and PA Community,

We are excited to inform you that our HERO (HElp Reduce Organic Acids) clinical study has begun.

The HERO Study is enrolling at least 12 patients with MMA and PA aged 2 and older at select leading children’s hospitals in the United States.

The study is currently open at the University of Minnesota in Minneapolis, MN and Children’s Mercy Hospital in Kansas City, MO. More sites will open soon and you can learn more about the study and locations on Clinicaltrials.gov.

The HERO study will assess HST5040, an oral therapy developed by HemoShear to reduce the toxins that cause harm in MMA and PA patients. HST5040 has the potential to be active throughout the body, including the brain, heart, liver, kidneys and muscles. The drug can be taken conveniently at home as a daily liquid formulation by mouth or through a gastric feeding tube.

ATTEND WEBINAR

HemoShear is collaborating with OAA and PAF to conduct a webinar, New Horizons for MMA and PA on Saturday April 17, 2021 from 1 pm – 2:30 pm Eastern.

This webinar will feature a panel of experts to educate families about the current state of treatment and potential future options. The speakers will talk about how the clinical research process works and share information about HST5040 and the design of the HERO Study.

us02web.zoom.us/webinar/register/WN_23hrLYL0R_6o8DlfGaNldA

YOU ARE A HERO!

Making medical progress to improve the quality of life for MMA and PA patients is going to be a collaborative process between industry, clinical researchers and families. We invite you to visit our website and sign up if you want to receive updates from us.

We are humbled and excited to see if our drug can make a positive difference in patients’ lives.

On behalf of the entire HemoShear team, thank you for your interest and support.

Sincerely,

Brian Wamhoff, PhD
Co-Founder and Head of Innovation
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.

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