Hope everyone is having a wonderful, healthy summer! I am excited to announce that on April 19, 2017 the Organic Acidemia Association launched our Natural History Registry for all organic acidemias! The official press release is included in this issue of the newsletter. This registry is an important step in furthering research for our disorders. I encourage all to complete the survey questions for your child (or yourself) with an organic acidemia. The website is https://oaaregistry.iamrare.org/. We currently have 55 participants signed up. This registry is different from other studies you may have participated in (the NIH, etc.). OAA is fortunate to have been chosen for this registry by a program with NORD and the FDA. If you have questions regarding your participation in this registry – please let me know.

I have also included information in this issue on several OA related organizations that OAA collaborates with. Working together is absolutely the key to give awareness and further mutual goals of our organizations. I look forward to working with these organizations on our mutual missions.

The next FOD/OAA Metabolic Conference is in the early planning stages! We will be hosting the next conference in Minneapolis, Minnesota next summer. No date or location has been set yet. We do have one major sponsor Mayo Medical Laboratories has agreed to sponsor our conference – however, we are looking for more sponsors – so let me know if you have any suggestions. I encourage all who are interested in helping to plan this conference to contact me ASAP.

My family participated this summer in the first study on Propionic Acidemia at the NIH. My Melissa wasn’t sure she wanted to participate – but ultimately we found out so much information on her and her Propionic Acidemia! There was so much information that I can’t hardly share it all here! I encourage all PA’s to participate in this study! Above is a photo of Dr. Chuck Venditti and his team with Melissa.

We are all in this together,
Kathy
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Organic Acidemia Association

OAANEWS.ORG

Launches Largest-Ever Study of Organic Acidemias

Research study is open to participants worldwide to advance understanding and treatments for rare organic acidemia disorders

GOLDEN VALLEY, MN  APRIL 19, 2017—The Organic Acidemia Association has launched the largest-ever study to research rare organic acid disorders that cause multiple life threatening conditions. Currently there are no cures for organic acidemia disorders.

“The OAA Natural History Patient Registry will provide a complete picture of each patient’s experience with a variety of different organic acidemias,” said Kathy Stagni, Executive Director, Organic Acidemia Association (“OAA”). “We are launching this initiative to help fill the missing link researchers and medical experts need to advance research, improve treatment and discover a cure.”

To help drive awareness and participation, the Organic Acidemia Association will contact families who initially showed interest at the OAA family conference last year. Plans are to post to social media (Facebook and Twitter) to invite families to participate.

“Our goal is to enroll as many patients, or their parents or legal guardians, as possible,” said Stagni. “The success of the registry is dependent upon community participation.”

The OAA Patient Registry is a natural history study that consists of electronic surveys to collect information about the patient experience and disease progression. Patients, or their caregivers or guardians, can enter information from anywhere in the world. The data are anonymous and stored securely in an online portal called a registry. The Organic Acidemia Association may share the data but not personal identifying information with individuals or institutions conducting research or clinical trials, subject to approval by the study’s governing board that includes scientists, doctors and patient advocates.

The Organic Acidemia Association is launching the study in collaboration with the National Organization for Rare Disorders (NORD), an independent charity that built a natural history study platform as part of its mission to help identify and treat all 7,000 rare diseases. Funding is supported by a cooperative agreement between NORD and the U.S. Food and Drug Administration (FDA). The FDA has praised NORD’s program as a helpful tool “that protects the security and privacy of personal information, while making valuable information available to a researcher or drug developer interested in creating a new therapy for a rare disease.”

NORD President and CEO Peter L. Saltonstall said, “NORD’s natural history studies platform empowers patients and families to drive research and eliminate some of the unknowns that still exist in rare diseases. We are glad to be working with our Member Organization on this project and thank the FDA for its support and ongoing commitment to help people with rare diseases.”

Organic Acidemias are a group of inheritable genetic metabolic disorders in which an essential enzyme that is necessary for protein metabolism is absent or malfunctioning. This defect results in a buildup of chemicals, usually acids, on one side of the metabolic blockage and a deficiency of vital chemicals on the other. This causes an over dosage of one chemical (often toxic) and a shortage of another which is essential to normal body functioning.

For more information, visit oaregistry.iamrare.org

About The Organic Acidemia Association (OAA) We are a volunteer 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.

About The National Organization for Rare Disorders (NORD) An independent 501(c)(3) nonprofit organization, NORD is the leading advocacy organization representing all patients and families affected by rare diseases in the U.S. Established in 1983, NORD is committed to the identification, treatment and cure of the 7,000 rare diseases that affect 30 million Americans, or 1 in every 10 people, through programs of advocacy, education, research, and patient/family services. In addition to educational resources for patients, families, medical professionals and students available on its website (www.rarediseases.org), NORD represents 250 member organizations and collaborates with many others in specific causes of importance to the rare disease patient community.

Contact: Kathy Stagni, Executive Director, 763-559-1797

¹Woodcock, J. “The more we know about rare diseases, the more likely we are to find safe and effective treatments.” FDAvoice (Oct. 23, 2014)
Introduction of the Nutrition Management Guidelines for Propionic Acidemia (PROP)

Overview
Nutrition Guidelines Project
The Nutrition Management Guideline for individuals with propionic acidemia (PROP) is part of a larger project undertaken by the Southeast Regional Newborn Screening and Genetics Collaborative (SERC) (HRSA Region 3) and Genetic Metabolic Dietitians International (GMDI) to develop nutrition management guidelines for inherited metabolic disorders (IMD).

Purpose
The Southeast Regional NBS & Genetics Collaborative (SERC) and Genetic Metabolic Dietitians International (GMDI) are proud to announce the Nutrition Management Guidelines for Propionic Acidemia (PROP). This has been a multi-year project to develop evidence and consensus based guidelines for nutrition management of inborn errors of metabolism (IEM). The PROP guidelines are now publicly available online to all metabolic dietitians, physicians and other clinicians.

Features
- The Management Guideline Portal is a tool for development of guidelines of genetic metabolic disorders for which there is little published scientific evidence.
- SERC and GMDI partnered to develop nutrition management guidelines based on our evidence and consensus DNDF1 methodology. The resulting PROP guidelines are freely available on this portal.
- Management Guidelines cover the assessment and management of patients known to have a particular metabolic disorder.
- The practice recommendations are an effort to increase standardization of care and enable outcomes studies within and across centers.
- When warranted by developments in PROP research and clinical practice, these guidelines will be updated periodically and will be maintained through a dynamic process.

Explore this resource at: https://southeastgenetics.org/ngp/guidelines-prop.php

Additional information available at: http://gmdi.org/Resources/Clinical-Practice-Tools/Nutrition-Guidelines

Many thanks to the contributors of this project which include the work group chairs and members, web developer, project manager, project coordinators, reference librarian, project consultant, project advisor, evidence analysts, dietitians, physicians, researchers, parents of children with PROP, adult patients, and reviewers. This collaborative effort has resulted in guidelines to improve the nutrition management of individuals living with IE Ms.

For more information contact:
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Dear OAA

This past fall we requested your help to understand your experience raising a child with an organic acidemia. The main purpose of our study was to identify the level of stress in parents caring for children with an organic acidemia. Moreover, we wished for parents to openly share their thoughts and experiences, and hopefully in turn, allow for identification of community and medical resources that may have a greater impact in lowering parental stress.

With your support, we had an overwhelming participation by parents all over the United States (and a few internationally!). We presented the findings of this project at the American College of Medical Genetics and Genomics on March 23rd, and we wanted to share our results with you as well.

For this study we used a questionnaire tool to measure stress as well as asked a survey of questions about how parents gathered information and support. It will come to no surprise to you that while the average US adult has an average stress score of 13.02; parents of children with organic acidemia had significantly higher average score of 19.4. Most parents (80.5%) reported moderate to severe levels of stress.

Parents with high stress more likely to report career decisions that involve not working outside the home. We think that parents who stay at home may be more stressed because their children are sicker and require them to be at home, but our survey didn’t really address this question. We also found that parents who were comfortable with the medical team and felt well prepared making medical decisions for their child had lower stress levels. So we think working to find a medical team you are comfortable with is important, but we know can be difficult given how few and far between metabolic providers are.

We are happy to find that participation in organizations like the OAA, were associated with significantly lower stress levels. Parents who received their information from flyers/brochures were found to have significantly higher stress levels. We believe the connection to other parents who have been in your shoes and are going through similar experiences has more of a positive impact than paper forms of sharing information. So we encourage you and your organization to keep up the great work supporting each other!

Overall, the survey findings revealed a significant number of parents of children with an organic academia have higher levels of stress than the general population, and organic acidemia organizations were identified as a key resource utilized by parents with low stress.

Thank you to the OAA for your support and participation in research of the psychosocial impact of metabolic disorders!

Sincerely,

Dr. Randeep Brar
Dr. Gabrielle Geddes
Medical College of Wisconsin
In April I was able to attend the Southeast Genetics Consumer Toolkit Meeting. A group of dieticians have been working on writing protocol for dietary purposes for a variety of metabolic conditions, including PA. The purpose of this meeting was to get feedback from parents on what they had written. Once we discussed our concerns we broke out into groups based on the metabolic condition we were representing. From there we took what they had written and re-wrote it into more patient and parent friendly terms. We did this mainly for parents who were newer to the disorder, so that they would understand what they needed to be aware of. We were able to brainstorm many ideas and the dieticians who were in charge of PA guidelines are in the process of re-writing these and getting a final draft. Once it is written and finalized it will be posted on the internet for easy access. This was a wonderful experience and it was great to see so much effort being put into making sure that parents are being provided the tools they need to help their children grow adequately.

AMBER, MOM TO SEBASTIAN AND GRANT, PROPIONIC ACIDEMIA

UPDATE:

Rhutu: MMA, Mut 0 | Age 22

Hi everyone here is an update about me. I graduated culinary school in New York in April 2014 as a pastry chef. October 2014, I got married to the most amazing, caring man ever. I am blessed. May 2015, my beautiful daughter was born natural delivery no complications and she doesn’t have MMA. I’m so happy, but her father is not carrier so that gave me better chances. I am currently working a full time job in NY in Times Square and my daughter is in daycare and is two years old now.

If anyone wants to reach out or contact me please email me rhutu32645433@gmail.com. I don’t have social media websites and I might take long to reply but I always reply, I just work a lot.

HCU Network America is a recognized US based 501(c)(3) non-profit organization dedicated to helping patients with Homocystinuria (HCU) and related disorders manage their disease and find a cure. HCU Network America strives to create connections, provide educational resources, and drive research towards a cure! We represent CBS deficiency, MTHFR and select Cobalamin deficiencies (Cobalamin C, D, J, F, G, E, D2) and some other remethylation defects.

What is Homocystinuria?

Homocystinuria is elevation of the amino acid, homocysteine (protein building block coming from our diet) in our urine. Homocysteine can also be elevated in blood. Untreated Homocystinuria can lead to blood clots in the lungs, brain, and other areas.

To learn more about Homocystinuria, please visit: HCUNetworkAmerica.org
Fishing for New Drugs for Propionic Acidemia

BY: VIRGINIA GINOCCHIO
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In the last decades careful clinical and pharmacological management have significantly improved the clinical outcomes in patients with propionic acidemia. However, the morbidity of the disease remains high and more effective therapies are highly needed.

We have generated a fish model of propionic acidemia using the Japanese medaka fish (Oryzias latipes; shown in the figure) as a tool for better understanding disease pathogenesis and for investigation of novel therapies. Fishes have been largely used to investigate developmental processes but more recently, they have also shown to be effective in modeling human metabolic diseases. Compared to other animal models used in biomedical research, such as rodents, fishes have several advantages including easy handling, availability in large number, and low cost. Moreover, fishes are well suited for high-throughput drug screening. These features make them excellent tools for investigation of novel therapies.

By genome editing technology, we introduced deleterious mutations in the fish gene that encodes for the enzyme deficient in propionic acidemia. Although they appeared healthy at hatching, these fishes showed progressive locomotor deficiency, lethargy and premature death in the days after hatching. Importantly, affected fishes showed the biochemical hallmarks of propionic acidemia, including increased C3 levels and the presence of methylcitrate. Like in humans, the administration of a low-protein diet and carnitine reduced the severity of the disease in the fishes. In conclusion, this novel animal model recapitulates several features observed in humans with propionic acidemia and it has potential to unravel the mechanisms underlying disease pathogenesis and to identify new therapies.

ASSOCIAZIONE ITALIANA Acidemia Metilmalonica Con Omocistinunria CBLC - ONLUS VIALE TITO LABIENO, 36 - 00174 ROMA – CF: 97926850583

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

The founders, coming from all over Italy, knew each other on the web, often the first shelter, after a first moment of suffering and shock, for those who come across a disease with a strange and complicated name for the first time. Day by day it will become more and more familiar and written several times into the search engines, hoping to find answers to questions that none of them ever imagined having to ask and to get in contact with people facing the same problems and fears.

In February 2013, after having met other parents and realized that she was not alone, a mother found the courage to create a virtual “family”: a group in which people involved in cbLC problems could recognize themselves, share experiences and help each other as much as possible.

More or less at the same time, a trial of an experimental drug involving a large number of families from all over Italy was started at the “Bambino Gesù” child hospital in Rome: this event gave the chance to transform many virtual friendships into real ones.

In 2014 such “family” was so numerous that the idea of creating a specific association for the cbLC began to take shape. Such step, ambitious and too demanding for many, remained just a good intention for a while, until the wide participation to the first data collection project for statistical purposes within the group members rekindled hope and determination once again.

Some years later, in March 2017, thanks to the stubbornness of five families, the cbLC Onlus was finally set up and is rapidly growing. In Italy several associations dealing with metabolic diseases already existed, but none with a specific focus on cbLC, notwithstanding Italy represents one of the country with the highest number of cases.

We are still very young but we have in mind a lot of ideas and projects that we are willing to share with all the interested stakeholders, both in Italy and abroad.

For further info and update, check out our web site www.cblc.it, currently only in Italian (English version to come shortly).
In 1991 my husband and I were so excited to find out we were going to be parents! Almost from the beginning though, things always seemed just a little bit off. During my pregnancy the baby rarely moved, yet I always had good checkups with the doctor. I had a bad feeling that I couldn't shake through the entire pregnancy, yet everything seemed fine on the ultrasounds and all my prenatal tests came back normal. I must have just been an extra nervous ‘mom to be’.

When our son Shane was born, he was long (nearly 22 inches) and 6 pounds 12 ounces. He looked really skinny, but other than that seemed perfect! All my worries disappeared, at least, for a short time. Shortly after birth little things started to show up; nothing major, just little differences. My best friend had a baby three weeks after I did. I remember her little boy, at the age of two months, already looking like a fat, happy baby. Shane still seemed like a newborn. He was floppy still, and wasn't growing as quickly. I mentioned it to the doctor and he told me to stop comparing my child with someone else's, all babies were different. Maybe he was right, I needed to relax. As Shane grew, it got harder and harder to not worry. He cried all day, every day. The doctor told us he had colic. “Just wait until he’s six months or so, he’ll grow out of it.” So, we waited. By the time he was one year old he hadn't grown out of it. On top of that there were new things to worry about. He was behind in everything he did. Sitting up, rolling over, walking, and talking. Not way behind, just….late. The doctor said “Give him time. Everyone does things at their own pace.” Ok, so we waited. He was tiny. He had fallen off the growth charts. The doctors told us not to worry. He'd catch up. So, we waited. He began to get sick a lot. He would run very high fevers every time he'd get a cold or an ear infection. The doctors told us he'd outgrow the fevers. So, we waited. At about the age of two he developed strange discolorations on his skin. His fingertips were dark brown, as well as his stomach. He had a large dark brown spot on his inner thigh, one on his inner arm, and his feet were dark brown near the toes. These spots were just birth marks. “Nothing to worry about. As he gets older they would probably fade”. So, we waited. All through this time I couldn't shake the feeling that something was wrong. None of my friends’ babies seemed to have all these little oddities. I knew I shouldn't compare. I’d been told that by every doctor we’d seen. I really tried not to, but as everyone else’s babies were talking and running, mine still only said simple words and walked using his hands to hold onto walls to steady himself. Still, he wasn't way behind, just enough to make me worry constantly.

Somehow he made it to age three. He was no longer crying all day, and he was talking and finally starting to run! Maybe he really was ok after all! In the meantime, we'd been able to give him a little brother, Kyler. Kyler was a fat, happy baby who didn't seem to have all the little oddities that Shane had had. Things were going good! Then Shane got an ear infection. On Halloween night we took him into Urgent Care to have it checked. He was wearing his little lion Halloween costume! He looked so cute, but so miserable. The doctor on call at Urgent Care was kind and caring. After confirming the ear infection, he asked me about Shane’s skin spots. I told him what we had been told in the past about them. He referred us to Dermatology just to have them looked at. What a relief! Maybe I wasn't crazy and someone was going to tell us what those spots were! The dermatologist we saw was very businesslike and not super talkative. After looking at Shane's spots, he said “Shane has something called Neurofibromatosis. Don't look it up. It will only worry you. I'll check him again in 6 months.” I went home and promptly looked it up. It terrified me. I called the kind doctor we had seen in Urgent Care. He brought us right in and looked at Shane's spots. He reassured me that they did not look like Neurofibromatosis, but they were unusual. He also noticed that Shane's ears were a little low set, he had fallen off the growth charts, and he was a bit behind in his development. With all these little differences, he thought it would be best to be seen by Genetics.

Now our journey was truly beginning! At age three and a half, Shane had his first genetics’ appointment. The doctor
was amazing. He diagnosed Shane with Noonan’s Syndrome, although Shane didn’t truly have all the features and some of his features that he had weren’t consistent with NS. It was the closest diagnosis they could think of. At that time there was no genetic test for it, but the doctor said it was coming. At some point they’d be able to test him for it. So, once again, we waited.

In the meantime our fat, happy Kyler had developed a strange lacy pattern around his neck. I kept thinking it was dirt caught up in his little neck folds, but I couldn’t clean it off. Then I began to notice brown spots on his arm, leg and more under his arm pits. He began to run extremely high fevers. He was even hospitalized for a fever of an unknown origin for over a week. His blood tests were coming back and they were seeing strange findings that they hadn’t seen before. They ruled out Cancer and Rheumatoid Arthritis. They ruled out Cat Scratch Fever and Asthma. No one could tell me what was wrong with Kyler. He didn’t seem to have the same features that Shane did, so they didn’t think he had Noonan’s Syndrome. He began to get better on his own, although he still, on occasion, would run extremely high fevers for a week or so at a time. He also began to slim down and eventually fell off the growth charts. Something was wrong with both my boys, but each seemed very different. Kyler hit all his milestones on time. He soared through kindergarten and first grade at the top of his class, while Shane had to repeat first grade and was given an I.E.P. Doctors thought maybe each boy had a different genetic condition, but my husband and I weren’t found to have any genetic concerns. How could both my babies have two different genetic diseases just by chance? The doctors at this point said they still didn’t know enough yet to give us definite answers. Someday they would. So, again, we waited.

During that time we had a little girl named Danica. She had none of the medical issues the boys had. As the kids grew, we saw Shane fall further and further behind. He was barely making it through school despite having an I.E.P and all the services we could get him. While he wasn’t running high fevers anymore, he would develop horrible mouth sores, and almost constant ear infections. He had to have several ear surgeries and eventually lost a major portion of his hearing in his left ear. At age 12, the long awaited blood tests that could diagnose Noonan’s Syndrome were available. Unbelievably, Shane tested negative. He did NOT have Noonan’s Syndrome. We were back at square one.

By age 14 he began having trouble walking. He’d trip a lot and began holding his arms out for balance. On top of that he had developed leg tremors. On to the Neurologist and many more tests. No one could find out why Shane was losing his ability to walk normally. He also seemed to be falling even further behind in school and we were told he would not graduate high school. He was referred to TRACE, a program in our area for young adults who have mild to moderate disabilities. TRACE would teach him skills to help him through life.

However, Kyler, despite a major health scare at age 11, (a ruptured appendix that kept him hospitalized for a week and a half), was flying though life. He excelled in school and was very involved in musical theatre, along with his sister Danica.

We were making our way through life and enjoying our awesome (yet sometimes unusual) kids, when we had our surprise baby, Kaden! What a shock little Kaden was, but he was a happy go lucky, mischievous little person and we were thrilled to have a baby in our house with all of our big kids!

Our biggest concern was Shane. He was such a kind and wonderful young man. It seemed so unfair that life was so hard for him and yet we had no answers. I was so involved with Shane’s issues that I hadn’t noticed (or, at least, hadn’t wanted to notice) that Baby Kaden was having problems of his own. He was born at a normal weight but after birth couldn’t seem to gain weight easily. We had trouble feeding him, and he was referred to neonatology although he was a full term baby. He slipped off the growth charts and doctors were concerned with his small size and feeding troubles. Like Shane had, he was developing slightly behind normal. However he was an extremely happy baby who was very laid back (and in a family with three much older siblings, that was a good thing!). One morning when he was three, I was getting him out of the bath tub and my heart stopped momentarily. While drying him off I noticed several of those same suspicious brown spots had begun developing on his little tummy. He began running extremely high fevers, and he would have febrile seizures. He developed mouth sores like Shane had always had. His skin was spotty like Shane and Kyler’s skin. As he grew older, he too, began having trouble in school, and eventually was diagnosed with ADHD and learning disabilities.

It was happening again. We took Kaden to Genetics, but sadly our much loved geneticist was leaving our HMO, so they sent us to a new one. This began more years of doctor’s appointments and genetics counselors but still no answers. There was more poking, prodding, research, questions, yet we still had no answers.

Kyler began having problems of his own. He went from being a straight A student to barely passing classes in high school. We thought he was being lazy, but was there more to it than that? No one knew.

At age 23, Shane had a job working at a hospital cafeteria (Where he continues to work today). He loved the job and was very happy with his life but he was tired of being researched. He asked us to take a break. After much discussion, we agreed. We assumed it was something no one would ever find out. It seemed there was no treatment anyways. Kyler and Danica were both in college and doing well.

[ CONTINUED NEXT PAGE ]
We were dealing with Kaden’s ADD, school difficulties and small stature. Maybe it was time to stop waiting for Science to give us an answer and start enjoying life. So that’s what we did, for a while.

In June of 2016 I took Kaden into his pediatrician. I mentioned his struggles that year in academics at school. He also seemed weak and lacked muscle tone. Our pediatrician felt it was time to begin again with genetics (No, not again!!). He assured me that there were new diagnostic tests available, and maybe this would be the time when we’d actually get answers. Therefore, back on the roller coaster we went again. Well, why not?

So we went back to genetics. They decided since Shane was the one who appeared to be most severely affected, they would test him, using all the genetic tests available. Shane was tested in August 2016, and we went on with our lives. After all these years I didn’t expect them to find anything. In October we received an unexpected phone call from the doctor. Shane had been diagnosed with a rare disease called Cobalamin F Disease. My heart was pounding. I couldn’t believe we finally had our answer! Kyler and Kaden were also tested and we received confirmation in February, 2017, that they also had Cobalamin F. Finally, after 25 years of waiting, our answers had come!

The Cobalamin F diagnosis put us on a new roller-coaster. However, this has been a more positive ride! In April, 2017, our boys were given the opportunity to be seen by National Institute of Health in Bethesda Maryland. This has been an incredible time of learning about my boys’ health. It’s also been an amazing experience being part of research that will one day help other patients. The best part has been finding out that there is medication that will actually treat my boys, eliminate some of their symptoms, and help them to stay healthy. Shane, Kyler and Kaden all are now receiving daily Hydroxicobalamin injections. Their Homocysteine levels have gone from dangerously high to nearly normal! Their strange brown spots on their bodies are fading. Our family now has hope and we are all excited as we can now look towards the future!

TAMMI
SAN DIEGO, CA

While dealing with the daily challenges of caring for a loved one with a chronic medical condition like MMA, it is imperative that we have HOPE. This includes hope for a brighter future for our loved ones as we witness their daily struggles while attempting to live with some sense of normalcy. We understand all about these struggles with the loss of one child to MMA while another child battles daily with the unrelenting effects of the disease process. Our daughter, Alyssa, is currently waiting for a kidney transplant which we hope will occur in the coming months.

The best outcome we can hope for comes in the form of better treatments and a cure for MMA. For this, we thank God for the talented research team at the National Institutes of Health (led by Dr. Charles Venditti) and their corporate partners like Selecta Biosciences, Inc. as they seek to cure this disease (and even other OA’s). For those of you who have had the opportunity to visit NIH and interact with the team, you understand how fortunate and blessed we are for their expertise. This team truly cares about our loved ones.

One way our family has personally attempted to show our appreciation for the research team’s dedication to our cause is to support their efforts through fundraising and raising awareness. That is why we created Angels for Alyssa (www.angelsforalyssa.com) almost 10 years ago. For the past 8 years, Angels for Alyssa has been administering the MMA Research Fund at the request of OAA. With the help of a number of our MMA families and generous donors, we have provided over $875,000 to NIH through 2016. With the additional funds we raised so far in 2017, we are currently just $60,000 shy of hitting the $1,000,000 mark by year end!

We would not be approaching this milestone without the direct support of our extended MMA families. This includes all those families who hold fundraising events like concerts, dinners and golf tournaments, offer recurring donations, seek matching corporate donations, hold birthday parties with donations in lieu of gifts, request support on anniversaries of diagnosis, offer donations in honor of loved ones and even memorial donations (sadly). However great or small the support, we thank you for your help! But especially, thank you for the gift of hope for all of our families to have a brighter future without the challenges of MMA. God bless.

BONNIE & DENNY MEISEL
PARENTS OF ALYSSA (AGE 19) MUT 0 MMA

** OAA also has an agreement with the NIH and our OAA Research Fund also sends funds to Dr. Venditti’s research team.
In the Rome labs they are building new rooms and spaces, they are hiring new researchers and they are also planning to have an aquarium to go on with other fish models for other organic acidemias.

Now our two researchers (a neuropsychiatrist and a dietist) are following some patients after liver transplant to check the new diet, because the new guidelines of liver transplants suggest having the transplant when the patients are 2-3 years old and so they are following the patients up to see if they are ok.

In these ten years we funded research for 353,000 euros, we hired three researchers, now the first one, Dr. Diego Martinelli, has been definitely hired by the Bambin Gesù Hospital, after he completed the stage at NIH last year, and we leased a tandem mass.

We translated the acknowledgements letter that we sent to all our friends so you can share below:

Dear Friends,

It looks strange but this year we wrote our thanks to you some days before the Walking, because we didn't want to lose what was growing in our hearts.

Now we are updating it with some numbers and results. 1200 people completed the 10th Walking smiling but tired for the uphill finishing and we sold 5800 tickets, 750 of them directly at the start.

During these months, while we were preparing the calendar, that this year is the “gift” that we gave to all the walkers we noticed that in these 10 years we make our way together many times and looking to all the leaflets of the past events we remembered with love all the friends who helped us to make special all these events, many of them perhaps we didn’t meet from years, but we didn’t forget their friendship, their generosity and the helping hand that they gave us walking with us.

There is also another thought that we’d like to share with you and we also want to thank you for that:

all the events that we organized in these 10 years, particularly the Walking, allow us to celebrate Davide, Luca and Simone in a great way and we can give them back all the greetings that we couldn't give them personally in these 10 years and so every kiss, every hug, every smile, every word that we exchanged with all of you for us are a way to say to our children Happy Birthday, Merry Christmas, Congratulation for your Graduation…

We don’t know what our children would have become when they grew up, but we know that they would have spent their lives as a “gift” and purely with joy, the same joy that you had, despite the hot weather and the pain for the climbing… because we recognized Davide’s contagious smile in each of you and also in all that helped us before, during and after the Walking.

A special thanks to the CoCoDa, the Committee of Davide Classmates: They never forget to celebrate their Friend: you all met them as volunteers at the start, at the refreshments tables in the woods, at the arrival and during the award ceremony… someone walked with all of you…10 years have passed but this friendship will never end.

We date all of you for next year and we hope we could transfer to you all of our gratitude, we still don’t know how much we raised during the event summing up donations, sponsors and tickets, but just like any year, we assure you all that the love, affection, closeness and friendship that you showed us yesterday were immense. Thank you from our heart!
Congratulations Malik; you did it!

Malik graduated from Franklin High in Seattle his senior year 2017.

“School years weren’t always easy for me; I was in the hospital a lot when I was younger and had a couple of hospital stays during high school and many absentees because I was sick. Every time I was absent or in the hospital I got behind and it was hard to catch up because I had to stay on top of my current classes. I found out in the last quarter of my senior year that I had 2 classes I was failing and I needed these credits to graduate. The teachers gave me the option of doing all the catch up online. I worked hard to finish and I did it; I graduated.

I am planning on attending community college this fall; I am going to start out with a couple classes to get used to it and then take more classes in the winter quarter. I am interested in studying videography. Its weird leaving high school but I hope I can find a job and study at the same time.

My diet is pretty restricted. I have been off formula for a little over 2 years. My protein intake is between 16 and 20 grams a day. It’s hard to get it all in because I am really picky. My mom and nana keep telling me I need to eat more vegetables and salad. I do eat them when they make them but I don’t make them for myself. I have to get better at that. My main foods are french fries, hash browns, low-protein pastas, rice (low-protein rice and regular mixed) and fruits. I have to remember to eat sometimes because food isn’t that important to me all the time. I drink ½ cup of milk a day instead of formula. I take L-carnitine (15 milligrams) and Vitamin D daily.

I used to think about my IVA a lot when I was younger and hate having it; and then having Sickle Cell too, it was just too much. I remember going to PKU camp and the only one there with PKU was my counselor. It was summer so everyone went swimming and I sat and watched because I can’t go into lakes because of the Sickle Cell. Any time I have been in a cold lake; I have had a Sickle Cell crisis. I didn’t like that camp at all. As I got older, I didn’t think about it as much it didn’t bother me as much.

I started going to Sickle Cell camp when I was about 11 and went every year until I turned 18. Everyone there has Sickle Cell so it was way better and I didn’t feel so out of place. I have never told anyone at school about the IVA and thru my senior project on Sickle Cell, I did tell my class I had it. People would ask me questions about my diet; “why I couldn’t eat meat” and why I went to the hospital, I just ignored them. It’s just hard being different and to have two life-threatening diseases is hard. My mom and nana keep telling me to tell people so they will understand. My family and a couple friends know; maybe someday I won’t feel so secretive about it.”

Mom’s Notes:

Malik was born and we thought he was a healthy baby boy until he stopped thriving at about 5 days old. Washington State wasn’t screening for IVA at that time so his life didn’t begin on a very pleasant note. He endured a coma and near death until he was finally diagnosed. It has been a journey; we are fortunate to have a great medical team, family and friends that have supported us along the way. Malik’s IVA was so chronic at the beginning that his diagnosis with Sickle Beta-Thalassemia (SCD) didn’t register until we finally realized how that would affect his life too. My daughter (D’Anya) has Sickle Beta-Thalassemia as well so I guess all I’ve known as a mom is taking care of special needs children. They are my blessings, my life and today they are both doing great. I have to continue to teach them about self-care as they get older and it’s all gone by so fast that I have to remind myself that they are becoming young adults.

I think about how life would be if they didn’t have these genetic diseases and I find myself thinking that I want them healthy but I love them so much just the way they are.

Written by Peggy H. (aka: nana), Seattle, WA

All content is from an interview/discussion we all had. I tried to represent their (Malik and Mom’s) words and thoughts as best I could.
D-2HGA is a very rare neuro-metabolic disorder in which cells produce a toxin that causes progressive damage to the cerebrum, the part of the brain that controls speech, memory, and movement. Since its discovery in 1980, there have been approximately 150 individuals diagnosed worldwide. Currently there are 31 gene mutations that can cause D2HGA, and two cases with no known gene mutation. Symptoms of D-2HGA include: Intellectual Disability, Muscular Hypotonia, Encephalitis, Seizures, Dysphasia, and Death. In some cases, there are no symptoms at all.

We are a family of five living in a small Kansas town. We have three children: Connor age 6, Nathan age 5, and Alyssa age 3. Connor and Nathan have both been diagnosed with D-2HGA, and even though they both have the same genetic mutation, they are on different ends of the spectrum when it comes to the disease. Connor has seizures which are controlled by one seizure medication, Nathan on the other hand has intractable epilepsy and is currently on four different seizure medications and has a VNS to help keep the seizures under control. Recently Nathan had pneumonia and had an NG tube placed because he developed dysphagia, and has lost the ability to swallow water without aspirating it. Despite their current health concerns, both boys are still very active, and on warm summer days can be found in the swimming pool or playing at the park.

I feel it’s important to share the backstory of how we got to this diagnosis because we did not find out from a newborn screening. I don’t remember the exact dates but I can tell you it took almost three years to get the proper diagnosis. Our journey started when Nathan, my youngest son, had a seizure when he was two. Doctors decided it must have been fever induced because he had been sick, but his fever had never been very high. Shortly after, Nathan started having what some health care providers called “abnormal movements”, he started twitching his head, and falling for no reason. For two years, these abnormal movements were spontaneous occurrences in our lives, which happened more when the season changed or when he was sick.

Connor: Age 6 D2HGA
Nathan: Age 5 | D2HGA

Every time we went to the hospital for help with these “abnormal movements”, I would advocate for my son saying there is something wrong with his body, and in response I was told there is nothing wrong, that they didn’t see anything, or that I was overreacting. I took video after video of his “abnormal movements” to show health care providers. When these movements occurred during an ER visit at a children’s hospital I was told, “he won’t get hurt falling like that”. But he was getting hurt from his falls, and there was no explanation for the unusual movements that were causing his falls. Finally, after two years of struggling to get help, KU Medical center heard my pleas for help for my son. Nathan was admitted, and after an EEG we were told Nathan had epilepsy. At last, progress. Unfortunately, that was just the beginning.

Nathan was placed on a seizure medication which made his seizures worse. I tried telling the doctors about this new development, and even though we had a neurologist, I could not get anyone to listen. Nathan’s seizures intensified to the point he could no longer function by himself. He could not stand more than a moment without falling. He couldn’t walk, talk, or feed himself. I called his neurologist office for help daily; I even called the after-hours number to speak with the on-call neurologist hoping to get someone that would listen. I went to our local ER where a doctor told us to increase his medication, so we did, and Nathan got worse. After what seemed like an eternity of watching my child go through hell, his neurologist got us in to a 48-hour epilepsy monitoring unit. Within the first few minutes of Nathan being connected to the EEG, doctors and nurses ran into the room to stabilize him. At last Nathan got the help he needed to stop the seizures, but we were not prepared for the diagnosis we were about to receive.

When Nathan was seen at KU med center, the EEG showed a certain kind of seizure, Nathan was placed on a medication called Trileptal which should have helped but instead it made his seizures worse. I remember our Neurologist asking questions about Nathans development. Nathan reached all his milestones on time, and despite having uncontrolled seizures continued to thrive. Nathans Neurologist talked to us about organic acidemia that could be the cause of his seizures, when D-2-Hydroxy Glutaric Aciduria was first mentioned I did some brief research and then brushed it off. I thought, there is no way, this condition is so rare and Nathan has none of the symptoms other than seizures. We consented to the urine screen and blood test, but we let it go and did not give it another thought. That is until we got the phone call confirming our worst fear; Nathan has D-2-HGA.

My life stopped that day, and I felt myself die. My sweet boy who loved life and had been perfect was ripped away from me. I worried about him and what his life was going to be like. I was mad at myself; I fought so hard to get...
him help and ended up with a diagnosis that is just unfathomable. I could not understand how all this happened, why there was no indication on his newborn screening and why, why did this have to happen. I spent days crying trying to figure out what was going to happen to him. After some time processing what was going on, and trying to wrap our brains around this big, scary diagnosis, which has no cure, we had our other children tested. My oldest son Connor tested positive for D-2HGA, but our youngest child Alyssa did not.

I wanted to write about the real feelings of what it’s like getting the unbearable news, what I was thinking and how hard the fight is to get medical treatment because I know we can’t be the only family going through this. Now that we have this big scary diagnosis, some might think that getting help is easier, but it’s not. Sometimes it’s harder. No one in our area has ever treated D-2HGA and on Nathans bad days, our ER visits might lead to a life flight to Children’s Mercy, which is 120 miles away. Our local hospital could probably manage his symptoms, but due to his diagnosis it seems like most doctors are overwhelmed and unwilling take on the responsibility of treating him.

Now that we have the correct diagnosis and we have processed all the information, we try to embrace that Connor and Nathan are rare, and they have a genetic mutation that is unseen. To us that means the boys can write their own stories and the small amount of research that has been done on D-2HGA, with all its bad news does not apply to us. We embrace life; we make changes that help the boys. For example, instead of Nathan doing physical therapy, he does gymnastics. There are no rules when it comes to rare diagnoses, so we roll with what life throws at us.

To the other families who are fighting to get medical treatment, or have a child that received a big scary diagnosis. Don’t give up. Stay positive, know that your child is an individual and their body is different than that of other kids, with the same diagnosis. Take the time to grieve, and then embrace the new. Your child is special but they are not defined or limited by their diagnosis. Seek out community support so that your children know how loved and special they are. For us we have the Eagle Riders of Junction City who are amazing, they are conducting a poker run to raise funds to keep the boys in gymnastics, so my husband and I can focus on paying for medical treatment. Most importantly, know you’re not alone, and that you and your children are not defined by a label or diagnosis.

With Love and Encouragement,

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Pregnant mothers can count on one certainty: that a clear or “normal” 20-week prenatal anatomy ultrasound produces an involuntary sigh of relief. You find out that everything is, quite literally, in place. You are halfway to the finish line and could really use a reassuring pat on the back. Despite discovering that we had a two-vessel cord, my midwife assured me that all was as it should be. Most umbilical cords contain two arteries and one vein. Ours was missing one artery. We had multiple ultrasounds until birth to ensure that baby was growing well and that there were no signs of physical abnormalities due to this small hiccup.

And all continued to be well. In April 2010, after a normal and fairly comfortable pregnancy, my son Nikhil was born. He came barreling into the world weighing a healthy 8lbs. I had a planned homebirth that was attended by my trusted midwife. Labor and delivery was everything they said it would be: loud, painful, exhilarating, slightly chaotic, and totally amazing.

I will never forget when the newborn screening (NBS) was done. The midwife came to the house and explained the process before poking the sole of Nikhil’s foot. That tiny prick on the heel looked so invasive at the time. And he cried and cried. How could a skinny newborn heel produce so much blood? If that was my heel, I thought, that prick would be comparatively massive. Of course, all babies in Ontario, Canada - where I live - have this performed shortly after birth. What began in 1965 with Phenylketonuria (PKU) screening, eventually expanded...
to include 29 diseases, all of which are now tested by Newborn Screening Ontario (NSO) in Ottawa's Children's Hospital of Eastern Ontario (CHEO). I quickly got over the excitement of the heel prick and passed it off as one of the many miniature heartbreaks that would accompany motherhood.

Remember those first few weeks of the newborn days? Me neither. It could have been days or a week, but I do remember getting the call. I was informed that my son had screened positive for 3-Methylcrotonyl-CoA Carboxylase (3-MCC) deficiency. They told me not to worry; that screening positive does not mean that the child is confirmed to have said disease. But, we would both have to report to the hospital for further blood and urine testing. As I always do when curious, I turned to Google and researched the heck out of 3-MCC and newborn screening in general. I learned that 3-MCC is an inherited condition that prevents the body from properly breaking down leucine, an amino acid. Leucine is present in many foods, and is commonly found in animal products, such as eggs, dairy, and meat. The malfunction or absence of the enzyme that breaks down leucine can lead to a buildup of toxic substances in the body and can result in issues with growth, health, and learning. This buildup can also create a metabolic crisis that, if left untreated, can cause seizures, brain damage, coma, and death. I knew that medical literature was nothing if not thorough, and was only slightly alarmed. This was just a positive screen, after all, not a diagnosis. After reading about symptoms and balancing probabilities, I decided that heading to the hospital for further testing was a mere inconvenience and that everything would check out normal, just as it had with our little two-vessel umbilical cord issue.

Let’s be honest: having to catch a urine sample from a newborn in a hospital washroom is not enjoyable. Neither is witnessing blood being drawn from his tiny, seemingly fatless arm. The truth is, I made my husband keep Nikhil company in the phlebotomy lab because I couldn’t bear to watch. What I didn’t completely understand at the time, however, was why I also had to provide the same samples. But I did. I just really needed to get the “all clear” from doctors so we could proceed with our normal lives. Our normal, crazy, exhausted lives with a newborn son.

When I was contacted by CHEO sometime later, I got mixed news about our results. My son, thankfully, was negative for 3-MCC. I, on the other hand, tested positive. Were they sure? They assured me that they were. My son’s initial positive screen was transient; that is, it was a temporary state that resulted from my own blood presence in utero. The initial screen picked up my status, but enough time had passed by the second test for my son to rid himself of my blood. This was the reason for giving my own samples in the hospital that day. Since I had previously read about the metabolic condition in great detail, I was incredulous. I had never experienced any of the listed symptoms in a chronic sense during my childhood, such as irritability, extreme fatigue, vomiting, diarrhea, and hypotonia. My mother reported that as a baby, I had no issues feeding or growing, nor was I afflicted with major bouts of illness. In 1984, the year in which I was born, 3-MCC was not routinely screened for. It was not until 2006 that it was included in the Ontario-wide screening program. Though 3-MCC is estimated to occur in 1 in 50,000 births in Ontario alone, it is not known how many asymptomatic adults exist. We are out there – but most are ignorant to their conditions, understandably so in the male population.

Since learning this new information about myself, I had worried about potentially passing my disorder onto future children. Fortunately, 3-MCC is inherited in an autosomal recessive manner. I am not a carrier and, therefore, cannot pass it on to my children. Interestingly, I have followed a vegetarian diet for over 18 years, long before I learned of my disease. Whether or not this practice has kept me healthier than I otherwise would have been, we will never know.

When I was first approached to contribute to the Organic Acidemia Association newsletter, I struggled with a reason to do so. I have no symptoms with which to sympathize, no advice to give about navigating the health care system, and no tips in dealing with restrictive insurance company policies. The reality is that, were it not for my son, I would likely never have known about my 3-MCC. But, by connecting with others through the OAA, I have witnessed a community that fosters support amongst its members. I have comforted new mothers who are facing the uncertainty of a positive screen. If nothing else, I hope that my story provides some hope that having a metabolic condition does not necessarily mean that a lifetime of illness is ahead. The scores of unrecorded asymptomatic adults are reasons to believe that we are not living at the mercy of our genetics. It is my sincere hope that as more disorders are added to the newborn screen, we have a real opportunity to tailor treatment plans to individuals, adding quality to the lives of those with organic acidemias.

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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 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 & 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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- 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.

OAA Internet Google Group

OAA’s main mission is to empower families with knowledge about organic acidemias. If you would like to connect with other families who share the same or similar diagnoses, please join our private OAA Group. Visit the OAA News.org web site to sign up.

OAA is on Facebook - donations can be sent through our “Cause” Page, connection with other parents can be found through our private “OAA Group” and private “Fan” Page.