NON-PROFIT TAX ID # 48-1038050 **VOLUME:** XXXIII, **ISSUE:** 1

Organic Acidemia Association



OAA Newsletter • Family Stories • Family Matching • Family Conferences • Research Funds Awarded • National Advocate for Newborn Screening

Parent Support, Education & Awareness www.oaanews.org

A support group for families living with methylmalonic, propionic, isovaleric, and other organic acidemias

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February 29th was Rare Disease Day

Don't you love Spring time! Hopefully all of you enjoyed an uneventful winter season. I've heard of many cases where our OAA children ended up in the hospital due to illnesses. Hopefully spring time brings renewed health and happiness!

February 29th was Rare Disease Day – this year we had several opportunities where we were able to spread awareness about our disorders.

Thanks to Lillian, mom to Connor, Propionic Acidemia, who represented OAA at a Rare Disease Day event at Baylor College of Medicine in Houston, Texas.

Thanks to Raymonde, mom to Marc Antony, MMA Cbl C – we have a wonderful Rare Disease Day t-shirt design. OAA sold these t-shirts as a fundraiser and raised almost $600 that will help sponsor families attending our upcoming conference.

And last, but not least, we've been busy planning our upcoming family conference will be held in Westminister, Colorado on July 8&9. I have included our tentative agenda in this issue – and you'll see we have many great speakers/topics! I hope you will be able to attend to learn about upcoming research and network with many OAA and FOD families. Hope to see you in Denver!

IN THIS ISSUE

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Get Your 2016 OAA Calendar

Support OAA by purchasing your calendar today! The calendar and other items can be purchased from OAA's **CafePress Store** - *www.cafepress.com/organicacidemiaassociation*.

Thanks again to Raymonde, mom to Marc Antony, MMA Cbl C for creating our wonderful 2016 OAA calendar.

ROBERT – ISOVALERIC ACIDEMIA, AGE 24
DEARBORN HEIGHTS, MICHIGAN

Austin : Glutaric Acidemia, Type 1 | Age 7

Austin was born at 1:26 in the morning October 1st 2008 after 13 hours of labor. He was slightly premature at 34 weeks, but was a big strong guy at 7 lb. 10 oz. Upon his delivery the nurse informed us he had "wimpy white boy syndrome", meaning he was lazy and was going into respiratory distress and immediately took him to the NICU. Being first time parents at eighteen and twenty-one we were in shock, but relieved and humbled when we checked out of the hospital after just five days. We thought the tough road was behind us and were so excited for our little family and the future. However, 2 days later on a Saturday afternoon we received "the call" or actually in our case it was a voice mail telling me there was a problem with Austin's newborn screening and we needed to contact the lab at Scottish Rite Children's Hospital immediately. My husband, mother, Austin and myself called and immediately left to go down there, once arriving they collected blood and urine and told me someone would contact me in a day or two with the results and if Austin appeared lethargic or began vomiting to bring him back immediately.

On Monday Emory Genetics called and said Austin's labs were back and his levels were through the roof. They told me to immediately stop breast feeding, as I was passing excess protein to him and to only give him formula. They tried to end the call there and say that we had to be in there office the following day at 9:00 am, but me being the hard head that I am, made them tell me what was going on. The nurse told me Austin had Glutaric Acidemia Type-1, where his body doesn't make the necessary enzymes to digest protein causing them to build up and cause damage, and not to Google it. She informed me a team of doctors would sit me down, explain everything and answer any questions tomorrow. Being so hard headed I of course googled it immediately. I got Gumby syndrome, metabolic crisis, developmental delays, irreversible brain damage and premature death. I saw horrific pictures of helpless kids, but was confident the lab made a mistake, that this couldn't possibly be my child because I did everything right throughout my pregnancy. I lived in a bubble, as my husband put it, didn't eat the bad foods, didn't drink any caffeine, and took all my vitamins. I even put head phones on my belly and played classical music!

The following day me, Austin and my mom loaded up and went to Emory for the first of many appointments. It wasn't a mistake, we were there for over three hours just listening to doctor after doctor. Thankfully they wrote everything down for us, because I couldn't tell you what was said. Finally Dr. Singh, our savior amidst all the confusion, told me the only thing that I need to focus on was the formula. It would be life changing and saving. Austin took Glutarex-1 eight times a day for 15 months, every three hours whether he was asleep or not. Then he moved onto Glutarex-2, for six times, then four and now it is three. He calls it his "milk" and we told him God made him extra special and only a few people in the world get to drink it, makes him strong and keeps the doctors away. I truly believe this an important key to his success. Along with being proactive about physical, occupational, and speech therapy which he received two times a week until he was 4 ½ years old.

We went to Emory every month for six months, then every three months and now it's every six months getting blood drawn at every visit. Austin was also diagnosed with Hydrocephalus, excess fluid on the brain, at three months. At nine months, after multiple sedated CT and MRI scans, they placed a shunt down his right side to slow head growth and let his brain develop normally. Everything worked great until January 2015, when he had a malfunction and required two surgeries and two additional shunts. Austin now has a team of doctors, a Geneticists, Neurosurgeon, Neurologist, Ophthalmologist, and Neuropsychologist as well as a Physical therapist, who work together to ensure the best for him. He is extremely active with baseball, swimming, golf, basketball, hunting and fishing. He is in the first grade, but receives some support services with reading and math. He loves school and is very outgoing. People say that he is an old soul and has a contagious spirit.

My main reason for wanting to share this is that I know it can be tough and at times isolating. We spent a lot of time at the ER his first three years. My mom even sprayed people with Lysol. I got asked how I do it and why haven't I broke down. My answer is always, if he can do it and be ok then I can too. He's the toughest little man I know. I have stayed away from the "metabolic family" partly because he keeps me so busy but partly because I don't want to come across as insensitive or boastful about Austin's health and progress. It took a lot of hard work and sleepless nights of worrying at home and in the hospital, but I know how fortunate we were to have caught it so early and to be where we are today. I would like to include my email *Jess1113@aol.com* for anyone who has questions, concerns, or just needs a friend to confide, complain, vent, or understand the shoes you are in. I hope this little peak inside my son's life will give you some hope.

Thank you for letting me share,

JESSICA
GILLSVILLE, GA | JESS111389@AOL.COM

i carry your heart with me

i carry your heart with me(i carry it in
my heart)i am never without it(anywhere
i go you go,my dear;and whatever is done
by only me is your doing,my darling)

i fear

no fate(for you are my fate,my sweet)i want
no world(for beautiful you are my world,my true)
and it's you are whatever a moon has always meant
and whatever a sun will always sing is you
here is the deepest secret nobody knows
(here is the root of the root and the bud of the bud
and the sky of the sky of a tree called life; which grows
higher than soul can hope or mind can hide)
and this is the wonder that's keeping the stars apart
i carry your heart (i carry it in my heart)

E. E. Cummings

a study

Health-Related Quality of Life in Children and Families Affected by Methylmalonic Acidemia

Knowledge of the psychosocial impact of methylmalonic acidemia (MMA) on children and families is necessary in planning counseling and therapeutic intervention. Although clinical outcomes of MMA have been reported, research on its psychosocial impact is limited.

In recent years, liver transplantation (LT) and combined liver-kidney transplantation (LKT) have been used as treatment options for patients with MMA mut0 with the goal of preventing hyperammonemia and metabolic ketoacidosis. After LT, patients have demonstrated increased energy, muscle strength, mobility, and protein tolerance; however, neurologic damage, kidney disease, graft loss, and death as a result of acidosis and sepsis have also been described post-LT. Given the varied outcomes, LT is not considered curative and there are ongoing efforts to determine its most appropriate role in the management of children with MMA.

The goal of this study was therefore twofold: (1) to measure health related quality of life (HRQoL) in children and families affected by MMA using the Pediatric Quality of Life Inventory (PedsQLTM) parent version and free responses, and (2) to assess the impact of LT on HRQoL by comparing responses between LT and non-LT patients.

From November 2013 to February 2014, parents and caregivers of children with MMA were invited to participate in a survey through the Organic Acidemia Association (OAA), a MMA Facebook group, the Stanford Children's Hospital biochemical genetics clinic, and members of the Metab-L mailing list. To be eligible for the study, participants were required to be a parent/caregiver of a child between ages 2 and 18 years with a reported diagnosis of MMA mut0.

A total of 35 responses from parents/caregivers of children (16 males, 19 females, mean age= 8.0 years) with a reported diagnosis of MMA mut0 were received. 71.4% of responses were from English-speaking parents/caregivers and 28.6% were from Japanese-speaking parents/caregivers. 45.7% of responses were from parents/caregivers of children with LT (7 males, 9 females, mean age= 8.4 years). Mean age at time of diagnosis was less than one year and mean age at time of transplant was 3.3 years. Of the post-transplant sample, 62.5% of respondents reported that their child underwent only LT and 31.3% reported that their child underwent LKT. 25.0% of respondents reported post-transplant complications.

Parents/caregivers were asked to select the MMA related interventions that their child receives. 75% of post-LT respondents reported that their child requires feeding by G-tube, whereas only 42.1% of non-LT respondents reported G-tube feedings. 31.3% of post-transplant respondents reported that their child requires hemodialysis, whereas only 5.3% of non-transplant respondents reported the use of hemodialysis.

Parents/caregivers reported lower scores on the majority of the PedsQLTM scales as compared to samples of healthy children, children with solid organ transplants for indications other than MMA, and families affected by chronic conditions. Scores for children with MMA were lowest in school and social functioning. Scores for families were lowest in worry and activity impairment. In free responses, parents/caregivers expressed that worry and impairment of daily activities, like family social activities, were caused by the potential for metabolic decompensation.

Liver transplant is not considered curative and there are ongoing efforts to determine its most appropriate role in the management of children with MMA

[CONTINUED NEXT PAGE]

Health-Related Quality of Life in Children and Families Affected by Methylmalon Acidemia continued...

Recurring concepts in parent/caregiver free responses about the psychosocial impact of MMA on the family included a better perspective on life, increased family member anxiety, stronger family bonds, social isolation, strains on family relationships, a general negative impact on the parents and child, and financial burden.

No significant differences were observed when scores from the PedsQLTM scales were compared between children with and without LT.

However, when asked, "In your opinion, did the transplant positively or negatively affect your child's development," the majority (93%) of parents/ caregivers of children post-LT responded that the transplant had had a positive impact on their child's development. Six recurring concepts about the impact of the transplant were identified from these responses, including health-related improvement, increased family social involvement, decrease in parental anxiety, a more positive outlook, and intellectual and emotional improvements in the child.

The majority of parents/caregivers reported positive impacts of LT in free responses. This positive perception of quality of life post-LT is similar to results from other studies of children with MMA post-LT. The discrepancy between scores on standardized HRQoL modules and parent reports may reflect greater disease severity in children post-LT as compared to children without LT. Children post-LT may have had more severe symptoms pre-transplant, leading to the decision for transplantation. The children with LT had a higher reported use of G-tube feeding and dialysis, suggesting that this group may have had more severe symptoms that necessitated these interventions. To control for these possible differences in disease severity, it would be helpful to administer standardized HRQoL modules before and after LT to the same patient/ family in future studies.

In summary, this study was the first to query parents/ caregivers regarding the impact of both MMA and LT on the HRQoL of their children and families using standard measures and presents the largest sample of children with MMA post-LT published to date. The results show a negative impact of MMA mut0 on patient and family HRQoL. In particular, school functioning and social interactions are impaired in children and daily activities, like family activities and household tasks, are impaired in families. There were no significant differences in LT and non-LT patient scores on the standardized HRQoL scales, yet the majority of parents/ caregivers reported positive impacts of LT in free responses. Findings such as these from HRQoL studies should be combined with developmental and medical outcome measures when considering LT as a treatment for children with MMA mut0.

"In your opinion, did the transplant positively or negatively affect your child's development," the majority (93%) of parents/ caregivers of children post-LT responded that the transplant had had a positive impact on their child's development.

We want to sincerely thank all of the families who participated in this project, and the Organic Acidemia Association for putting us in touch with many of those families.

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The Metabolic Diet App Suite:

Metabolic Diet Tracking on the Go!

Lifelong medical nutrition therapy (MNT) is a key part of medical therapy for many inherited amino- and organic acidopathies. But there is a lack of metabolic disorder specific diet intake tracking and meal planning resources. Many individuals living with metabolic disorders experience difficulties following their daily metabolic diet goals. The Metabolic Diet App Suite is a free, web-based metabolic diet tracking tool developed to help people living with metabolic disorders track and plan their daily diets. It was created with input from biochemical geneticists, metabolic dietitians, metabolic patient caregivers, and application developers for use on both mobile devices and desktop computers (*http://www.metabolicdietapp.org*).

General disorder information is provided for fifteen individual metabolic disorders for example 'GA1' (glutaric acidemia), 'IVA' (isovaleric acidemia), 'MMA' (methylmalonic acidemia), 'PROP' (propionic acidemia), as well as a general protein tracker 'PROT'. Food nutrient content is based on the MetabolicPro™ program food database, compiled by the Genetic Metabolic Dietitians International (GMDI) Technology committee.

Each Diet App offers functions such as: secured personal user login/password, user instructions, goal setting, daily intake tracking, food content checks, adding foods and homemade recipes, exportable daily food diary log (update your doctor and dietitian), and developer feedback. Individuals can start by going to the website and creating an account and diet profile, no downloading required. The website provides a user guide: 'How it works?' with answers to frequently asked questions (FAQs) such as how to recover forgotten password and usernames and contacting the developers. Initial pilot tester feedback was positive and suggestions used to further improve the App suite.

This project was funded by the British Columbia Children's Hospital Foundation (Treatable Intellectual Disability Endeavor in British Columbia, 1st Collaborative Area of Innovation) and the Rare Diseases Foundation (Vancouver, Canada). The Metabolic Diet App Suite is intended as a patient and caregiver support tool. They do not replace the GMDI Metabolic Pro™ diet analysis program or health care professional advice.

We hope that this tool helps ease the daily challenge of metabolic diet tracking and meal planning for individuals living with protein or amino- or organic acid restricted metabolic nutrition therapies. Ho G, Ueda, K, Houben RFA, Joa J, Giezen A, Cheng, B, van Karnebeek CDM. Metabolic Diet App Suite for inborn errors of amino acid metabolism. Molecular Genetics and Metabolism. 2016 Mar;117(3):322-7.

BY KEIKO UEDA MPH RD, GLORIA HO, RODERICK HOUBEN, JEFFREY JOA, ALETTE GIEZEN RD, BARBARA CHENG RD, CLARA VAN KARNEBEEK, MD, PHD, FCCMG

Our most beautiful day

The 6th of June 2015, the day we've been waiting for so long, was finally here; Our Wedding Day! We enjoyed that day so much, we'll try to make you enjoy our story of that day as much as we did. So, even when you are down at some point, you'll have to keep fighting for everything you love and all your dreams will come true, just like ours did.

Let us introduce ourselves first. Our names are Annemieke Vereecken and Marc Boënne (both 31 years old) and we both live with a metabolic disease. Annemieke has Diabetes type 1 from when she was 9 months old, over 30 years right now. It is one of the hardest forms of diabetes. She uses an insulin pump because otherwise she doesn't get her diabetes stable. Marc has MMA type B, non-vitamin B12 responsive. He was born with it and doctors first diagnosed him when he got in a Coma at the age of 2. With allot of pills and a strict diet we keep the disease under control.

A crazy combination, you'll think! That's what most of our relatives and friends thought. They were afraid that we were going for a very difficult future together. Because we have to be careful of what we eat which is different for each of us.

After a relationship of 10 years we can show them that we are the ideal combination and that we are happy. That doesn't mean we don't have hard times either. But it is easier for us to understand how it feels to have a disease, how it is to take medicines, how it is to live with a diet … An especially the diet is easier when you have to diet together than alone.

That makes our lives maybe a little more complicated that a normal couple's life, but we try to be as normal as we can be.

And yes you did read it correct, we are 10 years together and Annemieke had to wait that long for a proposal. It was 04/04/2014, the day of the proposal Annemieke got out of bed and she found a little poem from Marc. She is kind of spoiled on that area so it wasn't that abnormal. We ate breakfast together and we went to our jobs together. Marc first put Annemieke off at her job and then drove to his job. We don't work far from each other. It didn't take long before Annemieke got a strange call from reception, that there was a package for her and that she should come fast and pick it up. When she came at the reception desk Annemieke got a bouquet of 40 red roses. She was totally impressed. Annemieke saw the flowers came from Marc and texted him to thank him, but she didn't realized what was coming next. At the end of the workday, Marc normally picks up Annemieke right on time, but now he was late. She was waiting on the sidewalk, when an American Classic Car came rolling towards her and stopped in front of her.

Marc stepped out of the car, gave her one pink rose and said we were going to see a movie. Annemieke still had no idea.

Annemieke chose a movie in the movie theater and afterwards the American Old Timer picked us up again. But before he left, she had to open the trunk of the car. Annemieke found her own most beautiful clothes in that trunk, together with her shoes. She had to change.

With our finest clothes on, we drove to one of the best restaurants in Belgium, who prepared food that was adjusted to our diets. We enjoyed a fabulous meal and while we were dining Annemieke for the first time thought that Marc might propose.

But when we drove away with our one car from the restaurant to our home, she thought that it would never happen. When we came home, Annemieke went to the bedroom, which was full of candle lights and roseleaves. Marc told Annemieke to sit down and wait; he called "Belly", our Old English Bulldog, who came running up the stairs with a little package for Annemieke. Marc took the package opened it and asked if Annemieke would marry him. She said YES.

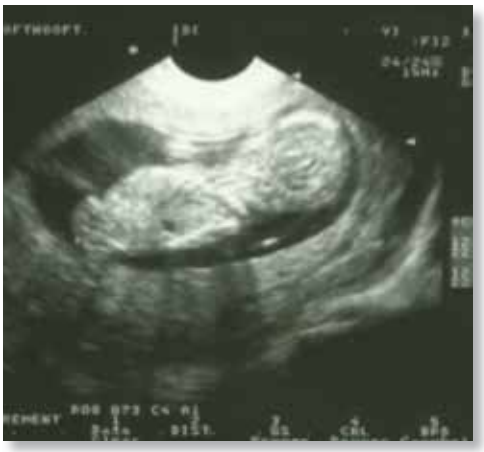
After that the big wedding preparations started.

Finding the right transportation, location, music and wedding clothes. We were busy with that every day until the wedding date 6th of June 2016.

We had to keep thinking about our diseases and diets when we were planning everything. Because we wanted to eat what we liked and were allowed to eat that day.

Finally the 6th of June the day had come…

The day began very nice with a nice open sky, with a lovely sun in it and 25°; (77°F) you can't have better weather than that in Belgium.



We slept separately the night before the wedding. So when Marc arrived with the family in a big shiny yellow bus, Annemieke and Marc couldn't be happier. It was really going to happened that day.

Marc rang at the door and Annemieke opened it. Marc gave her the bridal bouquet and she loved it.

Than the entire family came in and lifted the glass to the lovely couple. The entire family was in dress code, dark blue for the men, yellow for the ladies.

After raising the glass, we had breakfast all together and finally everybody got on the bus to city hall. At city hall there were already lots of our friends, who wouldn't miss this for the world. After signing the legal documents we were married and connected to each other for ever.

Afterwards we took some family pictures, because family is something important for us.

After the pictures were taken, we all got in the yellow bus again and went back to our home, where everybody enjoyed a nice lunch, adapted to our diet needs. We did this in our house so we could take our medication and take the rest of it in a bag for the evening party.

After lunch, the family stayed to rest in our home and the happy couple went to a wedding photo shoot in open air in beautiful weather.

When the photoshoot was ended we went to the garden of our wedding location, where we had a special wedding ceremony prepared where we had to say our vows and some friends said a few words. In a garden, beautiful weather, our closest friends and family (more than 200 people) and our ring carrier, Belly, our dog. It all made this the most beautiful day of our lives.

Many people shed a little tear during the ceremony; also for us it was very emotional. When the person who you love says he loves you in front of your 200 closest friends and relatives, it is something will never forget.

Time for the garden reception now! Also here there were adjusted snacks for MMA and diabetes. After the garden reception, people came inside for dinner. We chose a buffet, because everybody, including us, could take what they want and what they didn't want. There was also adjusted MMA and diabetes food. Everybody complimented us afterwards about the food.

At last, the opening dance, a very romantic moment for us, followed by a dance party that lasted until the early morning.

A day to never forget, to always remember!

Right now, we still think of that day sometimes, what a beautiful day it was.

And now we are ready for the next step in our lives, we are expecting our first child this summer and we are looking forward to it!

Before we met each other, we never thought this would be possible.

Because Annemieke needed diabetic values good enough to have healthy children, after a lot of effort, both us and doctors, it finally happened. Under strict supervision of doctors the pregnancy seems to be very good and without any problems. The idea of having our own child just makes us feel super happy.

This child will probably be healthier than the both of us.

Every day we are together is a party for us. We had to work real hard and fight for it (on medical domain), but no fight wasn't word fighting for if you see what we have right now.

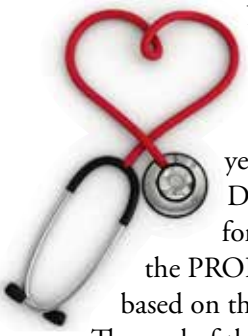
Fighting together makes you stronger than fighting alone!

That is why, dear people that we hope you had a positive vibe from reading our story. Don't give up your dreams! We didn't and we are still enjoying our lives and the world!

MARC & ANNEMIEKE
BELGIUM
EMAIL: MARC_FOR_FCB@HOTMAIL.BE

Are We There Yet?

Propionic Acidemia Nutrition Guidelines



It seems appropriate that the Propionic Acidemia (PROP) Nutrition Guidelines Nominal Group meeting in Atlanta, GA was held on a leap year, the weekend before 2016 Rare Disease Day. We have indeed leaped forward towards our goal of developing the PROP nutrition management guideline based on the feedback gained from the attendees. The goal of the Southeast Newborn Screening and Genetics Collaborative - Genetic Metabolic Dietitians International (GMDI) Nutrition Guidelines Project is: To develop nutrition guidelines for the management of genetic metabolic disorders for which there is little published scientific evidence. Evidence based medical practices should come from scientific findings gathered from randomized control trials (RCT) to guide clinical practice and treatments. RCTs require large numbers of participants to develop statistically significant conclusions, which is challenging to do with rare conditions such as PROP. Hence, most PROP publications are observational studies consisting of single cases or case series. In order to determine best Medical Nutrition Therapy for PROP, a group of experienced metabolic dietitians followed an evidence and consensus based methodology. The workgroup developed a multi-step process for guideline development known as the Delphi-Nominal Group-Delphi-Field Testing (DNDF) methodology1 , that includes a review of scientific and grey (unpublished) literature, a Delphi survey of practice, a nominal group meeting to clarify discrepancies, a formulation of recommendations and a second Delphi round to assess the degree of consensus with the proposed recommendations. External review and field testing are also built into the process. For PROP Nutrition Guideline, our workgroup of 65 individuals have been involved in some aspect of the development of the PROP guideline, including 13 experienced GMDI metabolic dietitians; have been trained in evidence analysis and DNDF methodology. We evaluated ------ 1156 articles, of which 225 published articles and 25 grey articles (book chapters, etc.) met the criteria for inclusion. The articles were rated for quality and summarized; the areas of certainty for PROP MNT and uncertainty requiring more clarification were identified. The nominal group meeting included the following metabolic experts; 2 physicians, 4 dietitians, 1 physician researcher, and 2 parent group representatives to participate in discussion and review of preliminary PROP nutrition recommendations. Topics addressed included acute and chronic management, energy and protein requirements, nutritional supplements and interventions to optimize outcomes in various circumstances such as pregnancy and transplantation, laboratory values to monitor, and much more. Our next step is to implement another Delphi survey to further validate the nutrition recommendations. The final step will be to summarize and write the PROP Nutrition Management Guideline that will be made publically available and submitted to a metabolic journal for publication by December 2016. We will periodically update them as new evidence to support best clinical practices are discovered. In the future a PROP tool kit will be developed for practical implementation of the PROP Nutrition Management Guideline, including educational resources. This project is supported by Maternal and Child Health Bureau HRSA grant #2-U22 MC010979. We greatly appreciate the contributions and support of the clinicians and the patient community to help us achieve this goal of improving MNT for people living with propionic acidemia, their families and caregivers.

PRINCIPLE INVESTIGATORS: RANI SINGH, PHD, RD AND FRAN ROHR, MS, RD

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UPDATE :

Stephen and Caroline :
Isovaleric Acidemia | Ages 18 & 13

I enter every spring with mixed emotions, because I am reminded of the day that life changed forever for us and Isovaleric Acidemia became a part of our lives. Our Stephen, the third of four children was one of many who received a diagnosis as a result of an acute metabolic acidosis that would nearly take his life, due to lack of comprehensive newborn screening when he was born. It's hard to believe that this May 30th will be fifteen years since that devastating day when our sweet 3 ½ year old son so full of life and possibilities would go from playing with his brothers to fighting for his life. He will forever remind us how life can change so abruptly when you least expect it. After three weeks in a coma on life support, Stephen opened his eyes and continued to recover, but time would reveal that his metabolic crisis would leave him with severe intellectual and developmental disabilities and multiple medical issues. In a way, we were the lucky ones, because so many families before us lost their babies and children in similar situations. After four weeks in the hospital and six weeks in a children's rehab center, we brought Stephen home to begin finding our new normalcy that revolved around his many needs and learning to understand him and how to care for him. We also had to adjust to the impact on our family and navigate our way through the changes as we each tried to heal from the tragedy that we all endured.

Eight months into Stephen's recovery, brought another life change. I was expecting another baby. Our daughter Caroline was born in September 2002. She too has isovaleric acidemia, but her diagnosis came early with an amniocentesis. Tom and I could not bear the thought of waiting until she was born to test for IVA for fear of her going into crisis after birth. While her early diagnosis was disappointing and saddening, we were able to process the news and come to an acceptance while preparing with the specialists for her birth. When it was time for her delivery, we were focused on just our baby girl coming into the world and not our baby with IVA. Since then, we have never let IVA define our daughter. Those early days after Stephen's crisis and Caroline's arrival seem like a lifetime ago, especially when I look at how much they have both grown.

Since my last update, Stephen has endured the many ill effects of his crisis. While remaining metabolically stable, his medical issues due to his neurological damage are his ongoing challenges. After years of progression of his scoliosis and kyphosis, Stephen required major spinal surgery for a spinal fusion. In January of 2011, he underwent a ten hour surgery to insert two titanium rods and twenty seven screws to straighten and fuse his spine. Due to multiple complications, Stephen spent thirty days inpatient. It was a time that once again, altered life for our family and reminded us of his days after his crisis. It was then that I called him my "apple cart." When you take one apple out, they all begin to fall out. When you alter one thing with Stephen, other things come unraveled. Despite the complexity and recovery, this surgery was worth every challenge, because it improved Stephen's quality of life immensely. He can finally hold his head up again independently after years of therapy. He can also tolerate and enjoy hours in his wheelchair when prior to surgery, he was not able to find comfort in his chair nor could he hold his head up. Time in his chair means more opportunities and outings, which he enjoys. His long recovery affected his positioning and allowed his hamstrings to tighten. One year later, he endured another surgery for bilateral hamstring release to straighten his legs again. He continued with a series of surgeries for issues that kept creeping up including a wound vac placement after the removal of a small deep tissue infection, the removal of his left testicle that retracted again and most recently, the extraction of all four wisdom teeth. While dealing with these issues, we discovered that Stephen was constantly developing a great deal of sediment in his urine to include small stones. After detailed testing, we discovered that he was consuming more calcium than his body needed given that he is not weight bearing. Together, the nephrologist and metabolic dietician worked to find the appropriate formula to meet his dietary needs while not overloading him with calcium or protein. Our challenge was finding the appropriate natural protein for him. He has been on Nestle Complete and Ivalex since then after removing milk from his diet. He was also diagnosed with neurogenic bladder, which was contributing to sediment build up and repeat UTIs. With this diagnosis, I learned my new skill of cathetering, because Stephen would need straight catheterizing multiple times a day to help empty his bladder. A few of Stephen's UTIs resulted in life threatening sepsis and time in ICU, but more frequent catheterizing and gentamycin irrigation inserted into his bladder three times a week has helped tremendously. He has done very well with this routine.

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[STEPHEN & CAROLINE
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Stephen continues to be our miracle boy and our little boy of so long ago turned eighteen this year! This was such a huge milestone to us that we decided to celebrate in a special way. As a family, we hosted the first annual Stephen Monaco Birthday Golf Outing and Dinner Fundraiser. It was a huge success and given how much we value the need for patient and family support, we decided to create a special fund at Children's National Health System in DC where Stephen and Caroline are followed. This fund will benefit the Division of Metabolism and Genetics and support educational programs and resources for patients and families along with research studies. We felt this was the best way to honor Stephen, since we couldn't celebrate in the typical fashion for an eighteen year old and it was a great way to bring family and friends along with staff from Children's together to celebrate him.

Turning eighteen brought another inevitable process our way that comes with having a child with special needs. We had to apply for guardianship for him, which was a long and grueling process, but a very important one. Once that was complete, we applied for SSI for Stephen to receive disability benefits since he was over eighteen. This was a more daunting process compared to guardianship, but again, one in Stephen's best interest and one he is entitle to have.

As we dealt with Stephen's issues over the years, we still had Caroline to manage. She has been quite healthy except for one admission for a gastrointestinal virus and the flu. Both required a five day hospital admission. Otherwise, Caroline's greatest challenge is gaining weight and consuming enough protein. She has consistently dropped on the growth chart for weight and is currently extremely low. She has never been a big eater, but thankfully, loves her formula. When she has been sick, it's the only thing I can rely on to sustain her through the illness. Aside from her medical needs, Caroline has grown into a happy, healthy thirteen year old. She is in seventh grade this year and does well academically. Caroline took dance lessons since she was five years old and played softball for a few years. However, she hung up her dance shoes and cleats for riding pants and boots when she fell in love with horseback riding two years ago. Riding horses and spending time with them is her greatest passion and serves as a wonderful way to build physical strength, deal with anxieties when Stephen is hospitalized and instill great life lessons in her.

As Stephen and Caroline have grown up over the years, they continue to inspire me to pursue my advocacy efforts for newborn screening and rare diseases along with patient and family centered care initiatives. Representing OAA in

Washington and on the national level helps to raise awareness for our organic acidemias and the needs of our patient population. Inserting my voice for OAA at this level helps to impact health care and policy for our disorders and ensure patient centered outcomes as these babies and children continue to grow up and reach adulthood.

As I reflect on our journey that began almost fifteen years ago, the image of that playful, happy go lucky little boy remains in my heart and mind. This year is particularly bittersweet as I ponder on the many lost dreams and opportunities in what would have been Stephen's senior year of high school. We think about it, but don't dwell on it. Instead, we celebrate his life each day, the happiness that he feels and experiences with us and the way his life has helped to change newborn screening preventing other children from sharing his fate. We find great joy in watching Caroline grow up leading a normal life with IVA and like her two older brothers, looks out for Stephen and helps with him. We celebrate how our family found our new normal over the years in this ongoing journey, since IVA became a part of it and are most grateful for the families in OAA who have supported us throughout.

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Recently Jana, mom to Stephen and Caroline, IVA represented OAA with NORD at the American Medical Association Convention. Jana had the opportunity to speak to many medical students and share awareness about our rare metabolic disorders

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1. Koletzko B, Lien E, Agostoni C, et al. The roles of long-chain polyunsaturated fatty acids in pregnancy, lactation and infancy: review of current knowledge and consensus recommendations. J Perinat Med. 2008;36(1):5-14.

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INTERNATIONAL METABOLIC CONFERENCE

JULY 8 - 9, 2016

AGENDA

FOD GROUP

Friday, July 8, 2016 Room: Westminster 1

TIME	SPEAKER	CONF. RM
7:00 - 8:00 am	Continental Breakfast for Conference Registrants	
8:15 - 8:30 am	Introductions by Deb & Kathy Keynote Dr. Janet Thomas – Children's Hospital Colorado - [also kids room open – Gray's Peak 2nd floor]	
8:30 - 9:30 am	**Kathryn Chatfield, MD, PhD** University of Colorado School of Medicine **TOPIC:** Cardiac 101 and FODs	
9:30 - 10:30 am	**Johan Van Hove, MD, PhD** University of Colorado School of Medicine **TOPIC:** Anabolism: What happens when fasting?	
10:30 - 10:45 am	**Break**	
10:45 - 11:30 am	**Mark Korson, MD** Genetic Metabolic Center for Education **TOPIC:** FAOD Management Strategies at Home and in the Hospital: Keeping your child safe	
11:30 - 12:00 pm	**Laurie Bernstein, MS,RD,FADA,FAND** University of Colorado School of Medicine **TOPIC:** Facts about Fats (Benefits of Micronutrients and Essential Fats)	
12:00 - 1:00 pm	Lunch for Conference Registrants	
1:15 - 2:15 pm	**Jerry Vockley, MD, PhD** Children's Hospital of Pittsburgh **TOPIC:** New treatments for FAODs (MCAD, long chain disorders and GA2)	
2:15 - 3:15 pm	**Nicola Longo, MD, PhD** University of Utah School of Medicine **TOPIC:** Clinical Aspects of Carnitine transporter deficiency as one ages (and similarities to other FODs)	2pm - DNA talk with OAA & FOD kids/teens-room TBA - Cindy Freehauf,CG
3:15 - 3:30 pm	**Break**	
3:30 - 4:30 pm	**Stephen Kahler, MD** Arkansas Children's Hospital **TOPIC:** Autism and mitochondrial disorders	
4:30 - 5:15 pm	**Professional Panel --- Q&A with several of our speakers**	
5:00 pm	**Summary and THANK YOUs!**	

OAA GROUP

Friday, July 8, 2016 Room: Westminster 2

TIME	SPEAKER	CONF. RM
8:30 - 9:15 am	**Laurie Bernstein, MS,RD,FADA,FAND** University of Colorado School of Medicine **TOPIC:** Nutrition Education and Compliance	
9:15 - 10:00 am	**Nicola Longo, MD PhD** University of Utah School of Medicine **TOPIC:** Liver Transplant in MMA-PA	
10:00 - 10:15 am	**Break**	
10:15 - 11:00am	**Jan Kraus, MD PhD** University of Colorado School of Medicine **TOPIC:** Enzyme replacement therapy for propionic acidemia-early attempts	OAA Cooking demo - Limit 25 Room TBA
11:00 - 12:00 pm	**Jerry Vockley, MD, PhD** Children's Hospital of Pittsburgh **TOPIC:** Novel therapies for Organic Acidemias	
1:15 - 2:00 pm	**Kathryn Chatfield, MD,PhD** University of Colorado School of Medicine **TOPIC:** Cardiac 101 and OAs	OAA Cooking demo - Limit 25 Room TBA
2:00 - 2:45 pm	**Steve Goodman, MD** University of Colorado School of Medicine **TOPIC:** GA1 Research	2pm -DNA talk with OAA & FOD kids/teens-room TBA -Cindy Freehauf,CGC or Shannon Scrivner,CGC
2:45 - 3:00 pm	**Break**	
3:00 - 4:00 pm	**Chuck Venditti, MD, PhD, NIH** **TOPIC:** MMA Update Cbl X – share time with **Tamim Shaikh, PhD and Curtis Coughlin**	
4:00- 5:00 pm	**Emily McCourt, MD** University of Colorado School of Medicine **TOPIC:** Ocular disease in Cobalamin C	
5:00 pm	**Summary and THANK YOUs!**	

6:00--- 9pm	**RECEPTION FOR FAMILIES, PROFESSIONALS, AND SPEAKERS**	

INTERNATIONAL METABOLIC CONFERENCE

JULY 8 - 9, 2016

AGENDA

FOD and OAA SESSIONS

Saturday, July 9, 2016 **** Wear your FOD and OAA t-shirts****

TIME	SPEAKER	CONF. RM
7:00 - 8:00 am	Continental Breakfast for Conference Registrants	
8:00 - 8:15 am	Introductions by Kathy & Deb **Remember PICS at 930am!!** Kid's Activity Room --- Gray's Peak 2nd floor	Westminster 1 (FOD) and Westminster 2 (OAA)
8:15 - 9:30 am	**FOD: separate room – FOD Breakout sessions/networking by Disorder** **OAA: separate room - Panel of Affected OA's**	Westminster 1 Westminster 2
9:30 - 945 am	**Break --- PICTURE OF EACH Individual GROUP WITH YOUR T-SHIRTS!**	**Open Room Divider**
945 - 10:45 am	**JOINT Presentation – Mark Korson , MD** **Topic: Genetic Metabolic Center for Education**	Westminster 1 & 2 together
10:45 - 11:45 am	**JOINT Professional Panel – most of our Speakers**	Westminster 1 & 2 together
11:45 - 12	Ending ceremony and slideshow	Westminster 1 & 2 together
	See you in 2018!	

Organic Acidemia Association

9040 Duluth Street
Golden Valley, MN 55427

ANNUAL DONATION CHANGE OF ADDRESS

Please accept $_____ as our annual tax deductible donation to the Organic Acidemia Association.

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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.

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

Find us on: facebook

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.

Google

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 *OAANews.org* web site to sign up.