Organic Acidemia Association

NEWSLETTER

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Summer 2007 Volume XXIV, Issue 2

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Editorial

This issue of the OAA family newsletter will cover a variety of milestones among our OAA families. My family experienced a major milestone this summer with Melissa's high school graduation. It seems like the other day she was born, and going through all the hospitalizations and illnesses. I'm pleased to include an update on her in this issue and hope you enjoy reading about her accomplishments. I strive to include a variety of family stories in each issue of the newsletter - and I hope that you'll enjoy and learn from all our families, even the ones with the more rarer OA disorders. Organic Acidemias collectively include about 16 different disorders. I have always felt that we should all bond with all the OA disorders and not focus just on our own disorder. We can all learn from one another.

So far this year, our OAA families have suffered many losses. As in previous issues, we have included a memorial section and also on the OAA website. Please take a moment and pray for our families who have lost their precious babies. I always feel a numbing feeling after the news of the death of one of our OA kids. Consoling parents or grandparents after a death is something I'm not trained in, but feel I've had my share of experiences over the past 10 years of running OAA. Personally my family suffered a major loss this year with the death of my mother-in-law. Betty Lee Stagni was such a huge supporter of my husband and I during the early years of caring for Melissa. I've included a tribute to her to thank the donations given in her name for the OAA.

I'm happy to announce that we have a supporter for another combination OAA/FOD conference. Dr. Jerry Vockley and Lynne Wolfe at the University of Pittsburgh have offered to host our next family conference. We are working on dates and hotels for the Summer of 2008. I will be asking for volunteers again to help with the planning of the conference. If you haven't already joined the OAA listserv, I invite you to do so. All the latest information on the conference will be posted on the OAA listserv. Messages of support are also offered with over 200 OAA families on the list.

I will conclude by saying that life has been quite hectic with a graduate in the house this year. Our family is also making major changes and we will be downsizing our home and moving soon. I appreciate the love and support from all of you. OAA's next newsletter will be sent out after the first of next year. We need your stories....physicians, we need your articles. OAA will be unable to produce a newsletter without them. The OAA board will strive to produce a newsletter that you will be proud to read. Speaking of proud, I would like to acknowledge and thank Janice Boecker (mom to Kristin, PA) and Juan Lopez (dad to Gabriel, PA) for producing the recent "Research in Organic Acidemias" publication. It has been a huge success and was distributed at the recent Scientific Conference on MMA at the NIH. OAA has plans to produce another research newsletter sometime in 2008.

Peace be with you.

Kathy Stagni
The 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 501c3 non-profit corporation. Donations to the OAA are tax-deductible. OAA publishes a newsletter three times a year, hosts an internet-based list-serv for information exchange and maintains a website. These services are funded by donations from corporation and individual members. Annual membership donation of $25 (U.S.) 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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FDA Approves Carnitor® SF (Levocarnitine) Sugar-Free Oral Solution For Patients With Carnitine Deficiency

GAITHERSBURG, MD, June 22, 2007 – The Food and Drug Administration has announced the approval of Carnitor® SF (levocarnitine) Sugar-Free Oral Solution for the same indication approved for the current Carnitor® Oral Solution containing sugar. Carnitor® SF and Carnitor® Oral Solution are indicated to treat primary systemic carnitine deficiency and for acute and chronic treatment of patients with an inborn error of metabolism which results in a secondary carnitine deficiency.

Adverse events reported with Carnitor® use include nausea, vomiting, body odor, gastritis, and seizures. There were no contraindications or warnings. Please see full prescribing information.

Primary systemic carnitine deficiency is a very rare genetic disorder that typically presents in infants. Secondary carnitine deficiency may present in infants, children, and adults. Factors contributing to carnitine deficiency may include inborn errors of metabolism, Fanconi syndrome, chronic renal dialysis, carnitine-deficient diet, extreme prematurity, malabsorption, HIV infection or antiretroviral therapy, valproic acid (VPA) and a Ketogenic Diet. Symptoms of secondary carnitine deficiency may include cardiomyopathy, encephalopathy, muscle weakness, anemia, and fatigue.

Carnitor® SF, the sugar-free version of levocarnitine, is appropriate for patients with carnitine deficiency for whom a sugar-free option is desirable. This may include patients with diabetes or those who are on a Ketogenic Diet who need to limit sugar and carbohydrates.

"We are pleased to be able to offer a sugar-free version of Carnitor® for patients who are diabetic or those who are on a Ketogenic Diet, those who are intolerant or have sensitivities to sugar and develop carnitine deficiency," said Gregg Lapointe, Sigma-Tau Chief Operating Officer. "In keeping with Sigma-Tau’s commitment to rare diseases, we are pleased to provide this new option for this important group of patients.

Carnitine functions in the body as a carrier of fatty acids to the energy centers in muscles (mitochondria). A deficiency of carnitine, normally produced by the liver and kidneys, can result in extreme muscle weakness and other related symptoms.

"Previously there was nothing available for patients with carnitine deficiency who needed to limit sugar intake, especially children with diabetes or those on a Ketogenic Diet," said Dr. Darryl De Vivo, Associate Chairman (Neurology) for Pediatric Neurosciences at Columbia University Medical Center. "Carnitine deficiency is a debilitating illness, so it is reassuring to know that these patients can still treat their symptoms without complicating their condition with undue sugar intake."

Carnitor® SF (levocarnitine) Sugar-Free Oral Solution is the only U.S. FDA approved prescription sugar-free oral solution of levocarnitine available. The product is expected to be available June 22, 2007.

About Sigma-Tau Pharmaceuticals, Inc.

Sigma-Tau Pharmaceuticals, Inc. is a U.S. based, wholly owned subsidiary of the Sigma-Tau Group, and is dedicated solely to the global development and commercialization of medicines for patients with rare diseases. Sigma-Tau Pharmaceuticals, Inc. is based in Gaithersburg, Maryland.

Since 1989, the company’s products have been focused on rare diseases, kidney disease, and cancer. With more than 6,000 identified rare diseases that affect approximately 25 million patients in the United States, Sigma-Tau places its considerable scientific resources behind the discovery of compounds that benefit the few. The company has a substantial development program focused on transplant, cancer, inherited genetic disorders, malaria, as well as other areas of unmet medical need. For more information about the company, visit www.sigmatau.com.

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Mead Johnson’s Helping Hand Program

Mead Johnson has a heritage of providing infant nutrition to babies with special needs, which is why we are proud to offer the Mead Johnson Helping Hand for Special Kids Program™. Mead Johnson believes it is important for infants and children to receive the formulas they need for good growth and development, regardless of the families’ financial status. Our program allows the child’s physician to, in partnership with Mead Johnson, extend a helping hand to families who need a specialty formula or a metabolic formula but cannot afford it.

Over the last 15 years, Mead Johnson has assisted over 5,500 families with the Helping Hand program.

The Helping Hand program is coordinated by the child’s physician, who works with the local Mead Johnson representatives to determine eligibility.

Nutrition News

Try Cambrooke Foods’ new low protein blueberry scones, and cranberry scones. These are satisfying breakfast comfort food, delicious toasted and spread with your favorite low protein spread, and convenient enough to eat on-the-go or any time of the day. Toaster Topz, coming soon, our good-for-you breakfast cakes - also perfect for pick-up and go food.

Experience a taste from the South of the Border with our new savory Southwestern Biscuits, a lunch or dinner item. They make wonderful accompaniments to your favorite meals and paired with a salad, make a complete lunch.

New ready-made Primavera Pizza, a traditional cheese pizza with a light vegetable topping is new in our single serve pizza line. Medley Meals join our expanding line of ready meals and are available in two varieties. Heat and serve convenient, the preservative free Vegetable Masala and Moroccan Stew can be eaten as is or served over your favorite low protein pasta or rice. Traveling with a metabolic disorder will never be the same because these new meals are not only delicious, but they are shelf stable and nutritious!

Cambrooke Foods is ALWAYS open to serve you. Call toll-free, (866) 4 LOW PRO / (866) 456-9776 or visit our website at www.cambrookefoods.com. If this is not convenient, you can mail (2 Central Street, Framingham, MA 01701), e-mail (orders@cambrookefoods.com) or fax at (978) 443 -1318.

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**Upcoming Conferences/Socials/Fundraisers**

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<tr>
<th>Event</th>
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<tbody>
<tr>
<td>IOGA Summer Social</td>
<td>Friday, September 14, 2007</td>
<td>5:00PM at the Clinic for Special Children</td>
<td>Cay Welch – 724-459-0179</td>
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**Iota Club & Café and B.O.S. Music to Present 5th Annual Dave Daly Memorial Fund Benefit Concert**

B.O.S. Music, a next-generation record label based in the Washington, D.C. area, and Iota Club & Café in Arlington, Va., will team up on Saturday afternoon November 10, 2007, to host the fifth annual Dave Daly Memorial Fund Benefit Concert. The confirmed line-up for the show includes such artist’s as Philadelphia’s Ben Arnold and Boston’s cutting edge roots rock sensation Oneside (with more artists to be announced).

Dave Daly, who touched many people every day through his good nature, intelligence, charming wit, and limitless generosity of heart, passed away in 2002. The Dave Daly Memorial Fund was founded by family and friends in early 2003, with the goal of bringing light from darkness, and to keep Dave’s memory alive in a positive way. The Fund’s events, focusing on golf and music (two things Dave enjoyed), rely on volunteers so that the maximum portion of the proceeds can be donated to deserving charities, as well as fund annual high school scholarship awards presented in Dave’s name.

All proceeds from this year’s event will go to benefit the OAA/MMA Research Fund. "My father didn’t have MMA, but we’ve decided to start selecting deserving charities to benefit from our events, and I have a couple of very close friends who have a child with MMA," says Skip Daly, the primary organizer of the benefit concerts. "It’s a relatively rare condition, so it’s good to make an effort to bring some visibility to the cause."

MMA is a group of inherited genetic disorders that result in the build up of methylenalonic acid in the bloodstream. This build up can lead to a severe condition, which can be fatal for the affected patient. This disorder affects about 1 in 25,000 to 48,000 people. (This rate may be higher because some neonatal deaths may be attributed to undiagnosed metabolic disorders.) The OAA/MMA research fund was launched in early 2000, to support ongoing research. In the summer of 2003, research on MMA and its director moved to the National Institutes of Health (NIH) in Bethesda, MD, where work continues today with the addition of staff and support. The OAA/MMA Research Fund holds all donations throughout the year and issues grants to the MMA Research Project at the NIH semi-annually. 100% of the money donated to the OAA/MMA Research Fund goes to NIH’s research on MMA.

The 4th Annual Dave Daly Memorial Fund Benefit Concert
Saturday November 10th
2:00 PM - 5:00 PM Doors open – 2 PM
FEATURING: Ben Arnold • Oneside ...and more TBA
Iota Club & Café - 2832 Wilson Blvd. Arlington, VA 22201
703-522-8340
Donation/Cover: $15 (ALL AGES SHOW)
More Information: www.DaveDalyFund.org
www.MMAresearch.com

**Donations from Metabolism Families**
(reprinted with permission from the Family Newsletter at Children’s Hospital of Philadelphia)

The Raccosta Family and the Bennett Family both donated either money or gifts to our department and the Children’s Hospital of Philadelphia. These families were able to help over ten families during the holidays. Six year old Melanie Bennett (IVA) asked if she could make Holiday gift bags for other children at CHOP who would be in the hospital over the Christmas holidays. Melanie and her family passed out the gift bags to some of the children who were in the Oncology Day Hospital. She also left extra gift bags for other children. We all were overwhelmed that such a young person would be so empathetic to the needs of other children. Melanie truly showed what is truly the spirit of Christmas! Thank you so much for all you do!
Lawrenz Ewers, 3 years old, Propionic Acidemia

Laurenz was born on March 3, 2004, and is now three years old. He truly was the missing puzzle piece in our family. We also have a set of twins, Antonia and Kilian, who are six years old.

When Laurenz was born, he slept completely for two days straight, and had no appetite. In the first three days his body weight decreased 100g daily, and his body temperature steadily decreased 1°C each day. On the third day we decided to take him to a children's clinic, and if we hadn't done so he might have died a short time later.

Initial blood tests showed a metabolic disorder. His blood was then sent to Heidelberg, where Dr. Hofmann made the diagnosis. Laurenz had Propionic Acidemia, a disease we had never heard of.

When Laurenz was 5 days old he was transferred to a metabolic clinic in Munich. A constant infusion of glucose was used to remove the poison from his body. Three days went by before he finally opened his eyes again. After another three weeks he was released from the clinic, and we brought Laurenz home. He recovered relatively quickly, and soon developed normally according to his age standards. He slept through the night, and took his daily rations orally.

In August 2004, he began refusing food, and would shove the bottle out of his mouth whenever we attempted to feed him. My mother-in-law and I stayed by him day and night, feeding him as soon as he slept. In October 2004 we moved to Hannover, and introduced Laurenz to the staff at the Medical University of Hannover. At this point Laurenz was already in poor condition, losing weight and vomiting regularly. Three days later we drove Laurenz to the emergency room, after another bout of continuous vomiting. Unfortunately, despite the emergency plan that we had brought with us, the situation was falsely evaluated. Laurenz received his nutrients through a feeding tube – but he received the vital glucose infusion much too late. He suffered through seizures for about 12 hours. Since that tragic night, Laurenz has been severely handicapped. He cannot crawl, walk, or stand, and speaks only about 30 words. Since then Laurenz receives his nutrients through a PEG feeding tube because he has completely lost his sucking reflex.

Laurenz is full of joy and life. Despite his disabilities, he can still move around by scooting on his bottom. We currently undergo different types of therapies with him, in the hopes that he might one day regain the ability to stand, walk, speak, and even eat by himself. Caring for Laurenz is a labor of love, and we enjoy every day that we have with him. He has made our lives much richer, even when it’s sometimes more stressful and not always easy dealing with the jealousy of his siblings.

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Josh Mayfield, 23 years old, Glutaric Acidemia Type 1

Hi. My name is Josh Mayfield. I am 23 and I have GA1 also known as Glutaric Acidemia Type 1. I have had this from age 2 it is cause from to much protein. I am a member of OAA so I can get people to come to my site and I can help them out by emailing and chatting with them. I like to help people with disorders and things. I have met people with the OAA and I helped them get a bill passed for newborn testing in West Virginia. I tell about that on my site. I would really like to tell more about me. I like to talk on the phone, chat, work and go for walks. Now more about GA1 and things. I use to go to Pittsburgh to the Children's Hospital. I use to go there for doctor appointments. I don't how much, but it was a lot. I haven't been there for years now. So I have been doing good. I am off my low protein diet. I was on it for years, but the doctors said I could be taken off it. I graduated school on the honor roll in 2002. I went to West Virginia Division of Rehabilitation Center in 2004 and they kicked me out in 2005 or something like that. I forget what year, but why they kicked me out is because they were scared I would fall and get hurt and get a law suit. I still don't think that was right. But you can read more on my site about it. That's the end of my story. My website is http://www.jkmga1story.info/index.htm.

You can look at it and Email me at kickbutt162000@yahoo.com
Thanks your GA1 friend Josh

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My Brother
By: Melinda Bazzy
(sister to Robert, IVA)

My brother is different from all others,
He has a disease like no other.

He has had always enjoyed
different things, He even likes to sing.

I love him like a best friend,
More than any other type of friends.

Even though he is different from others,
He is still very special because he’s my brother.

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Stephanie (Age 8) & Jack (5 months) Divin, D-2-Hydroxyglutaric Aciduria, D2HGA

I’ve been meaning to write Stephanie’s story (and now Jack’s story) for a long time. It just seemed like such a monumental task. The recent death of a very special baby boy with D2HGA has inspired me to sit down and write our story. Some people think sick children are "less", not the perfect healthy child everybody prays for when they are pregnant. Our D2HGA kids and other sick kids are so much "more"; they are special, and selfless. They don’t judge, they don’t hurt others, they don’t want more than we can give, they teach us to appreciate the simple things. Trust me, I have 2 other "typical" children so I know how self centered a child can be! They are better, beyond that – a gift!

Our Daughter, Stephanie, was born on May 4, 1999. It was the day after our 3rd wedding anniversary and I couldn’t believe I had a girl! I felt so lucky. I had a son, Ben, who was born in 1997; he was almost 2 years old. What a perfect family! I always felt Stephanie was special; it was intuitive. I wondered why I felt that way, I had to appreciate every moment; I thought it was because she was a girl, what I always wanted.

Stephanie had jaundice when she was born, it was not a large amount and she never had to go under "the lights". I remember this because she slept a lot, and we couldn’t get her to open her eyes for the hospital baby picture. When I finally got the pictures in the mail, there was a poem about a sleeping beauty and how she wouldn’t open her eyes for the photo. I thought it was strange her jaundice didn’t go away until she was 5 weeks old; she had so little of it to start with. Stephanie had normal baby illnesses, colds and such. Our son was very healthy and never sick, so this was all new to me. When she was 8 months old she was baptized, during that time she was suffering from a fever for well over a week. During her baptism the fever went away, just for that morning, I felt God had His hand in this. It turned out she had a urinary tract infection. I had brought her to the doctor a few times, but the locum (our doctor was away) just gave me the same old story about "it happens with some babies, don’t worry". Finally when our doctor came back, he ordered a urine test for infection, it was positive! She suffered for 2 weeks with fever and pain for the stupidity of a locum doctor! I felt bad too, since I’m a laboratory technologist and should have asked for urinalysis.

When Stephanie was 2, she seemed to "stop" her progress with speech. Ben was very gifted and was reading by the age of 3 so we just thought we were comparing to him and really she wouldn’t develop as quickly as he did. At 3 Stephanie went to preschool, she still wasn’t toilet trained. I remember those phone calls where I’d have to go change her & take her home for doing "a poop" in her diaper. Nobody else’s child was wearing diapers. I was disappointed with her.

In the second half of preschool, as Stephanie was approaching her 4th birthday, she still wasn’t talking more than a few words. Speech therapy was suggested to us and we went. From there we were referred to the paediatrician who ordered many tests on Stephanie and said "this looks like low IQ". I remember those words like they were yesterday. Stephanie was also suffering from various infections ranging from strep throat to a possible blood infection; she seemed to always be sick. I got a call from the paediatrician in May, just before we left on an Alaska cruise, - Stephanie had just turned 4. She said there was something wrong with Stephanie’s chromosomes. It turns out she has an inversion of chromosome # 12 on the "p" arm. I took a picture of her in the yard that day, so I would remember the exact time we found out there was something wrong with Stephanie. I can’t look at that picture without crying. Later I found out I too have the same inversion, as does Jack and Martina (one of our healthy children). Whether this inversion has anything to do with D2HGA or if Stephanie has more going on there than we do, is yet to be determined. I thought that was all that was wrong & started researching everything about chromosome #12. A few days after we got back from our holiday I went to see the paediatrician. There was this other thing, an increased 2 hydroxyglutaric acid in her urine, but they’d have to send it off to Amsterdam in Holland for further testing to see if she had the D type or L type. The Paediatrician seemed not too concerned about it. It came back weeks later that Stephanie had D-2 hydroxyglutaric aciduria. They told me it was very rare, and at that time there were about 40 known cases worldwide, even now there are less than 100 known cases. There is no known treatment or no cure, such heavy words to hear. How was it that I was able to meet and marry a man who is a carrier of the same, unknown, defective gene? I can’t even imagine the odds of that! Soon after this all the appointments started. Stephanie also started taking CoEnzyme Q10, since starting it she is rarely sick and I feel it has really helped boost her immune system as well as helping minimize the effect of D2HGA. She also had to have baselines for all her organ systems so we could monitor the changes over time due to degeneration. It was such a tragedy for me to know everyday she will be a bit worse, that today she is healthier than tomorrow. As a mother it’s a hard thing to take.

During this time, and still today, I have become so frustrated with the doctors and specialists. I’ve always been one to want to know Stephanie’s test results as soon as they’re ready; it’s probably the "lab tech" in me. I felt the doctors were unwilling to share those results, they treated me like "I didn’t need to know that", when, in fact, I did. It would ease my mind to know what was going on with her. I could understand what was going on in her body and most of all it would help me to prepare questions for my next visit. There was one "glowing" exception, our geneticist, I could email her and ask questions, (continued on next page)
In June of 2004 we had another baby, a girl we named Martina. I was so afraid she would have D2HGA, but she didn’t. I was so happy to have her; she was my 3rd child. Stephanie, unlike other 5 year olds, was not interested in the baby at all. She didn’t want to help, or look at her. I had to force her to be in a photo with her. I was accustomed to her “D2” ways and accepted this. We had just had her psychological assessment done and her IQ was borderline for mental retardation. In some parts she was at a 1 year old level and she was 5 years old.

Stephanie started kindergarten in a private Christian school in the fall of 2004. I remember my biggest fear was she would "run away" during playground time, she had no sense of danger. She would go with anyone, and she had no fear. I remember I lost her in the supermarket one time, I was frantic and very scared; when I found her she was very calm in the "bulk food" section eating from the bins. She could care less that her mommy was missing.

In spring of 2005 Stephanie was diagnosed with ASD, (autism spectrum disorder). This explained a lot about her, why she always played by herself, why she wasn’t ticklish, why she put everything in her mouth, why she would twist her arms and run way when people spoke to her, and so much more. Since then I have learned so many children with metabolic and mitochondrial disorders have autism (the defective enzyme for D2HGA functions in the mitochondria). I think every child with autism should be tested for those diseases. The diagnosis of autism also allowed for more funding and "help" for Stephanie in grade one, the following fall.

In spring of 2006 we went on a trip to Maui, Hawaii. It was a great family vacation and Stephanie acted almost "normal". We had so much fun, but I was sick for the last week of vacation. In spring of 2006 we went on a trip to Maui, Hawaii. It was a great family vacation and Stephanie acted almost "normal". We had so much fun, but I was sick for the last week of vacation.

On a Thursday, when Jack had just turned 2 weeks old, there was a message on the answering machine to phone the geneticist. I was rushing to pick up Ben and Stephanie from school so I phoned her from the car on my drive to school. She told me "unfortunately Jack has increased 2-HGA." I couldn’t believe it, my sweet baby Jack was sick too. I said something brave like I knew it was possible, but I was so sad inside. I phoned my husband and told him, he didn’t react nearly enough for my liking (but he’s one to keep feelings to himself) and I just felt so alone, so sad for my 2 "D2 kids". When I told Stephanie, who liked her baby brother about as much as she liked Martina as a baby, she smiled and said "I’m not the only one". I don’t think she knows what D2HGA is but she knew she was the only one in our family to have it, so now she wasn’t alone.

Jack also had jaundice when he was born, he had such a small amount they didn’t even test him for it. He too took a very long time to clear the jaundice. He was yellow until 7 weeks old, and for starting out with such a low amount it seemed to take too long to clear from his body. He has most of his appointments ahead of him. He’s had many blood tests & I always feel bad for him when they hold him down and take his blood. He looks at me and cries. I can’t help him.

It is now June of 2007 and Jack is doing well, he has "low tone" to some degree, and mildly delayed physically, but otherwise he is doing well. I don’t know yet, how he will be mentally. I am so afraid he will develop autism, a large percentage of D2HGA kids have it. I am trying to spend so much time appreciating his smiles and interactions with me, because they may slowly fade away, we are making memories. He sleeps with me every night, I want him close. I want him to love me for as long as he will be before he too becomes autistic.

Stephanie is finishing grade 2, she has "teacher’s assistants" one on one with her and she likes them. We have trouble with her schoolwork, and homework is torture for us. Many times we’ve thought "why bother sending her to school" but she goes. She loves her brother Ben more than us. I don’t understand why since he’s so bossy to her. She questions why people do things; she doesn’t understand why people are mean. I think we can learn from her.

People have asked me "how do I do it?" They feel bad we have sick children, but I feel bad for them. It’s a whole different world we live in. I can’t even imagine if my biggest worry was whether my child failed a test or didn’t make the team. Stephanie and Jack have taught me to appreciate just that they live.

Outi Divin
Mom to Ben (10), Stephanie (8) D2HGA, Martina (3) & Jack (5months) D2HGA
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divin@shaw.ca

(continued from previous page)
Memorials

Mark Mansfield, MMA, Mut 0  

I meant to do my work today……  
*By Mark, December 2006*  
I meant to do my work today  
But I day-dreamed of sand and castles  
And saw a sky of blanket blue  
I meant to do my work today  
But I saw a bird warming her wings  
And a cat laying, lazing in the sun  
I meant to do my work today  
But a Samba Band made me dance  
Like a tree swaying in the breeze  
I meant to do my work today  
But a glassy river wound to the sea  
Through land, with lush, green grass

A memory from Christine  
- Mark’s sister  
There are so many memories. But to share one – it is symbolic of his fearlessness, his courage and his independence. Refusing my assistance at Steephill Cove, Mark’s determination to clamber over the slippery rocks alone, infuriated me. I was terrified that he would fall. Of course, he never did fall. I resigned myself to sitting and watching him on his solitary journey. He had a look of sheer concentration and utter peace in his eye, and was triumphant in his success. I’ll never forget that moment when I realised how much he embraced life – throwing caution to the wind.

Tricia Stackpole Stratton, D2HGA  
(D-2 Hydroxyglutaric Aciduria)  

God's Garden

God looked around his garden and He found an empty place.  
He then looked down upon this earth and saw your tired face.  
He put His arms around you and lifted you to rest.  
God’s garden must be beautiful, He always takes the best.  
He knew that you were suffering. He knew that you were in pain.  
He knew that you would never get well on earth again.  
He saw the road was getting rough and the hills were hard to climb,  
So He closed your weary eyelids, and whispered “Peace Be Thine”,  
It broke our hearts to lose you, but you didn't go alone,  
For part of us went with you the day God called you home.

In Memory of, Betty Lee Stagni

My mother-in-law died on February 28, 2007 and I would like to pay tribute to a woman who was a constant source of support for me and our family. My daughter Melissa was the love of her life and even though we didn’t live nearby, she was a true advocate and was always interested in how she can help support Melissa. Thank you to the following who made memorials to the Organic Acidemia Association and **OAA/PA Research Fund** in Betty Lee’s name – Kathy Stagni -

Alvin & Judith Babeaux, Thibodaux, LA 70301  
Rosalie Marcello, Thibodaux, LA 70301  
Ann Barker, Terrebonne Motors, Houma, LA 70360  
Michael Barker, Barker Mitsubishi, Houma, LA 70360  
Mrs. Philip Ayo, Thibodaux, LA 70301  
R. & G. Lewis, Thibodaux, LA 70301  
Delta Coin Machines, Inc., Thibodaux, LA 70301  
James Mayon, Thibodaux, LA 70301  
Steve & Stacy Finkelstein, Plymouth, MN  
Cindy & Tony Winiarski, Agawam, MA  
Co-Workers of Kathy Stagni from UCare Minnesota, Minneapolis, MN

Thank you for the generous donation to the OAA/PA Research Fund made by Lorraine Osowski in memory of her granddaughter Kathy. Lorraine and the Osowski family have been great supporters of newborn screening and in promoting research to find a cure for PA.
Adrian Lopez, Age 4, 3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)

Adrian, our first and only child, was born in March, 1st 2003. He weighted 7 pounds 15 ounces and was 20, 1/4" inches height. He was a healthy normal baby. He scored 10-10-9 APGAR.

By the end of the first month, Adrian developed a very high fever and nothing seems to work. We tried a bunch of oral antibiotics but neither one worked. Adrian lost weight and didn’t get over it. He was decayed. After several tries, we decided to switch doctor and the new one detected a urinary infection and decided to treat it aggressively with an intra muscular antibiotic. For 10 days, Adrian received a shot in the morning and another at night. It hurts more Lorena and myself to watch our little son receive his injections without complain, just a small moan. Luckily the treatment worked very well, and Adrian recovered positively.

While he was growing we realized that he was not developing like the other children. He has a cousin that was born one week before him and was doing so much things that he did not. With 7 months he did not seat by himself, it could not raise its head and did not roll over; in addition he did not have strength in his muscles. We began to ask his doctor and she said us that it was normal and that each baby develops at his own steps.

In a visit to another doctor, she worried much about some reflex and other nervous stimulations that did to him and advise to us to visit a neurologist. We visit the neurologist and he ordered a tomography and an encephalogram. We did not do the tomography because we were scared of the anesthesia. The results that he told us did not convinced us. It is just that feeling that something is wrong but you just don’t know.

In the beginning of 2004, we hire a physiotherapist to help him achieve his milestones. He noticed that something wasn’t right with his right hip. We took Adrian to get an x-ray and then to a trauma doctor and he confirmed that he had DDH, (developmental dysphasia of the hip), a common issue with new born and easily treated if caught in time (at three month of age). Of course that wasn’t our case. So we start looking for a children traumatologist doctor. We found that finding one wasn’t easy. I think there are only three in Santa Cruz. His surgery took place January 21st and supposed to be a success. He had to be in a cast for 90 days. After 4 weeks in a control x-ray the hip was out of the hip socket and back to the surgical bed for correction. After two weeks in another control, the hip seemed to be out again. This time we took a tomography to confirm the situation of the hip. Also we used this anesthe-

sia to do the head tomography. Anyway, the hip was out again so the cast was out.

After this, we didn’t know what to do, so we went to his neurologist to see what he had to say about the tomography. He just looked at it and said it was OK. Moms, and grandmas, instincts kicked again and took him to another neurologist in Cochabamba, a city about 40 minutes fly, to double check it. This one scared the Jesus out of us. We were concern about the hip and he told us that he had Cerebral Palsy. Say what? We cried a lot… we were devastated. He told us that he couldn’t be with a cast for to long, that he had to do so much work to minimize the CP. Back in home we start to search for treatments. In a family meeting an aunt told us about this hospital in Argentina, Hospital Universitario Austral, witch had very good doctors. As a young couple with a small child, we didn’t have the resources to go there, but with the help from ours families and from this aunt, we took Adrian for a full study.

So in April, we went to Argentina for hip and CP appointments. Once there the doctors check him and with a MRI ruled out CP, that was a relief. But it wasn’t all right. He had some damage someplace in the brain that had affected his motor skills. We asked them what to do with his hip and explain to them what the other doctor told us. They recommended to "fix" the hip ASAP so this wouldn’t interfere with his developmental milestones. One of the doctors thought it was recommendable to do some metabolic test to rule out a metabolic disorder. By that time, we didn’t have any ideas about metabolic disorders. We took Adrian, to "Laboratorio Dr. N.A. Chamoles" and leave the urine and blood samples. This is the only lab that does this kind of tests in all Argentina, and one of the few in South America.

We went back to Santa Cruz – Bolivia, the city we live a little more calm, unaware of the A-bomb that was going to be drop on us.

A few weeks later we received a call from Dr. Chamoles telling us that something was wrong with our baby. “Get Albicar (L-canitine in Argentina) and give him 1g a day for 30 days and then send us some more urine and blood to redo the labs. Don’t feed him with proteins. Your son has an Organic Acidemia know as 3MCC” he told us. We moved the earth to get this drug and started with "treatment".

With this little information I jumped to the Internet to find out what was this thing that our baby had. We found some technical information (you may need a degree to understand it), some devastating stories and some not to scary ones. One of the sites I went thru was OAAANews.org and they had a mail list that really help us a lot. By that time we didn’t know too much about OA’s and had so many questions. Some of them were answered by the mail list member and especially with "plain English". That prepared us a little more and we had an idea of what to ask the doctors.

After the 30 days we sent the samples and waited for the results to come out. With the results we fly back to Argentina for new appointments with the doctors. With a little more knowledge, the doctors explained to us what 3MCC was and how was going to be the treatment. The also arranged an
We started to make the arrangement and back to Argentina.

Six month after diagnosis, he was stable and ready for surgery. He gained weight. His diet and medicines were very controlled. He recovered very well.

Changed. We kept him with therapy for motor skills and with a metabolic nutritionist that explained Adrian's diet and the drugs he was going to use. By that time Adrian was 1 year 2 months; he didn't walk, and was about 22 pound. 1.5 g/kilogram of protein, 1g of carnitine, 1,2 g of glycine.

About the hip, they told us that he had to be metabolically stable to perform a surgery.

After knowing what was the main issue with Adrian, things changed. We kept him with therapy for motor skills and with a very controlled diet and medicines. He recovered very well. He gained weight.

Six month after diagnosis, he was stable and ready for surgery. We started to make the arrangement and back to Argentina. The surgery took place in October 2004 and was a success. Adrian handled the surgery pretty well, and stayed in the hospital for 3 days only. One of the most shocking moments was when I took my little boy to the surgery board. They want me to take him so he would be calmer. I felt his fear when he graved me in a big and strong hug. Then the anesthesia went in and he felt sleep, but didn't release me. I felt an enormous emptiness. I still remember it like if it was yesterday. A few weeks after, we returned home with Adrian in a cast.

Two months past by, with all the troubles that is having a child with cast and finally the cast was removed December 22nd, almost one year after all begun. He cried a lot but he calmed very soon. We went to the apartment and he started crawling. That was our Christmas miracle for us. But we knew that our job wasn't finished yet, we had a long way to go.

Adrian finally walked a few months after he turned 2 years. He is metabolically stable and by now is a normal child. The hip is going regular. Maybe he will need a new surgery in the near future.

Now that I can think a little be more clearly, I realize that all this events, the hip, the CP, etc., lead us to truly find the real reason our baby wasn't developing like he should. If the hip wasn't wrong we maybe wouldn't go to the doctors and particularly to that neurologist that told us that he had CP. These really shook our world and got us to Argentina and to a final diagnosis and treatment.

I have to thanks our FAMILY, especially Lorena's mom that traveled with us to Argentina and stayed with them for a couple of weeks, the doctors in Argentina, Dra. Grañana, Dr. Amantino, Dr. Albarracin, Dra. Marchione; his doctor in Bolivia Dr. Arias; and the people that supported us in many ways, Carol and Kathy.

Thank you very much.

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Audrey "Audie" Gail Spegal MMA
August 19, 2001 - July 1, 2006

Audrey "Audie" Gail Spegal was just shy of turning 5 and starting pre school when she past away July 1st 2006 from a disease called MMA + HCU.

She loved the outdoors, animals and enjoyed watching John Wayne movies. She was a huge fan of Broadway musicals; her personal favorites were Oklahoma and Singing in the Rain. (I can still see her dancing in front of the house in the rain singing the theme song). She lived, loved and laughed every single day with a smile on her angelic face.

First, let me say as a parent I have never heard of MMA much less HCU until the day came we had to find out what our healthy daughter died from. It was a Wednesday morning and she did not want to get up out of bed and was complaining of an upset stomach. I just thought that she ate too much at her sister’s ball game the night before. She said that her tummy hurt and she did look rather pale so my instincts told me to take her to the hospital immediately.

Once there, they did a blood test and found out that she was in renal failure and she needed to be transported to Riley Children’s Hospital for dialysis. This was the first shock to my world, as I did not know that she was even that sick at all. We were at the hospital only 12 hours and she was sent into surgery for dialysis. We were informed from the doctors there that through a stool sample they had found that she had gotten into E. coli and it was shutting her kidneys down.

The E. coli got in her system (we don't know how) and caused her to have HUS (Hemolytic Uremic Syndrome) The renal doctor assured us that she would be fine in time with the most drastic thing to worry about was dialysis and a kidney transplant. Ok with dialysis she could live a normal life later or so we thought. After 28 days on dialysis in ICU everything that was being done stopped working. She was retaining fluids faster than the dialysis could take off and the antibiotics seemed to be given in vain. Her condition had gotten worse. She now had the HUS with the E. coli being treated but also peritonitis, pancreatitis; her blood acid levels were off the charts and a staff infection along with now fighting off blood pressure problems. By day 29 the doctors where stumped and did not know what else to do after 6 blood transfusions no matter what they did the blood acid level was very high so they began to investigate the possibility of genetics after (continued on next page)
We put her on life support after she went into a coma and exhausting every other test.

We were told that she had two rare genetic metabolic disorders as well as HCU.

Our 4-year daughter who had a bright future and was never sicker than the common flu was now gone and I am now leaving the hospital without her. There is never a more empty feeling as to leave a hospital without your child. I now knew what it would have been like to lose her at birth. The total and unforgiving emptiness was haunting. I felt cheated and empty more so than I had ever felt in my life. Her death certificate read that she died of HUS and we were still shocked by the outcome and we had unanswered questions that loomed over us. "Why, if it was not fatal, did it take her?" "Was there anything else that could have been done?"

Looking back everything medically possible was done to fix the acidosis in her blood. We consulted with the top Renal and Cardiac doctors there and they used every bit of knowledge they had. All of the nursing staff became very attached to my brave Audie and when she passed there were a few that could not finish their shifts and had to leave because of their attachment to her. She was very brave and stoic about all of the treatments that were given. They saw this special child clear to the end coming to the funeral to say their final goodbyes. The nursing staff at Riley is the best I have ever seen and they did everything they could to keep her happy every day. They even sang to her and painted her nails those nurses are a rare breed. It took 2 weeks for us to get a call from the Genetic Lab at Riley Hospital and we sat down with Dr. Hainline and his staff.

We were told that she had two rare genetic metabolic diseases, MMA + HCU. (Update: It has been confirmed that Audie had MMA Cobalamin "G"). Not just one but two and she was asymptomatic on both. We were shocked that she was born with this and we didn't even know it.

What is even rarer is that my husband and I are both carriers of the disease and we didn't even know it. Dr. Hainline informed us that every person is born a carrier of at least 5 genetic diseases without showing any symptoms or signs. It's when you have a child with someone else that has similar genetic traces that causes the genetic problems. Audrey received a recessive "Tainted" gene from each of us at birth. Each child had a 1 in 4 chance of being infected. That really is a scary thought to think what could have been by just mere changes. She was considered a rare atypical case where she had the disease at birth and did not have the side affects that went along with it. She should have had neurological difficulties as well as other side affects. She had nothing; I repeat nothing that even showed a glimmer of what took her!

The disease in her body lay dormant until the E. coli invaded her body and caused her immune system and white blood cells to go into overdrive. Thus unleashing the MMA which consequently began to take its course like wild-fire in her little body. She fought with all 38 lbs of her body and she was a brave soldier to the end never crying out and never complaining. Even when she was taken off solid food and all liquids for 28 days.

Her tissues were donated to help further the research for MMA+HCU and it is my hope that someday strides can be made in research and my precious daughter can be part of something positive in the OAA. MMA has many faces and different mutations, but not all are typical. She was a special case where being atypical is very rare. We had our other two children tested for MMA+HCU and found them negative however they do have a 50-50 chance of being a carrier. When my children are ready to have children they will have to have my grandchildren tested for MMA. I hope that the research has progressed in that amount of time for all types of Acidemia disorders as well as HCU.

My heart breaks for all of the parents who are reading this that have faced what I did or perhaps just finding out what their child is battling. Audie’s legacy has taught me to be strong and not to have fear because she was truly the bravest soul and I was blessed to have been her mother. She will always be our special angel.

I look forward to chatting on the OAA listserv and I know that I am not alone.

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Nikki Elizabeth Hrichak
Glutaric Aciduria Type 1

Ten years. One decade. It is amazing how much has changed in the world in some ways and how so little has in others.

Nikki was diagnosed with Glutaric Aciduria Type 1 10 years ago this week. She just celebrated her 10th birthday. She swam in the pool, jumped on the trampoline, broke the piñata, and threw water balloons, right aside of all her 4th grade friends. She brought home good grades and participated in dance, voice, tennis, and saxophone lessons. She won the citizenship award two years in a row. She generally sticks to a vegetarian diet (by choice) but does occasionally choose salmon, scallops, or shrimp for her main course. She suffers no apparent ill effects from small amounts of concentrated protein and we no longer treat her diet any different from her sisters.

She does take her carnitine and calcium pantothenate in her morning juice every morning and I do have to remind her that she has this “disorder” and needs to be thankful for each and every day. Due to the tireless work of people like Dr. Holmes Morton and the staff at the Clinic for Special Children, the members of OAA, Save Babies through Screening, and so many other organizations, Nikki does not have any memories of ever being “sick.” The only time we really notice a difference between our two girls is when Nikki gets a fever. It seems to affect her more than her sister and Nikki’s fevers only respond to Motrin.

For those of you who are not familiar with Nikki’s story, I will recap it. Nikki was born in June 1997 with a normal midwife-assisted delivery. She nursed well and seemed to be thriving just as her older sister had done. We opted to have the expanded newborn screening test taken as it was only 3 additional drops of blood and $25. On day 13, we received the call that would change our lives: Nikki had GA1 and needed immediate medical attention. We were referred to Dr. Morton at Lancaster General Hospital. Nikki’s diagnosis was confirmed and she was started on valium and L-carnitine. I was insisting that Nikki continue to be fed breast milk and Dr. Morton agreed if I would pump so we could measure her intake. I did-- for the next 17 months. Nikki never received formula. When I was no longer able to meet her needs, we weaned her to Rice Dream enriched which she drinks to this day. We counted every calorie and every gram of protein she ingested for the first 2 years of her life, keeping charts and records. She met every developmental milestone on time (except speech as her very verbose 3 year old sister spoke for her so there was no need.)

At 16 months, she had a set back with a subdural hemorrhage from a very minor fall. The neurologist at our local hospital (where I happen to work as a PT) intimated that it followed the signs of abuse; however, nothing came of that passing suggestion. We were lucky! Nikki was taken by Medivac to Lancaster and once again, Dr. Morton took part in a miracle. Her bleed extended while in the hospital, mimicking a stroke with hemiplegia. Dr. Morton used Decadron to help decrease the cranial inflammation and Nikki walked out of that hospital without any sign that she had a setback.

We have continued to visit Dr. Morton on a yearly basis. I think he continues to be amazed at the why and how Nikita came through all this without a trace of the effects of GA1 when so many other children, even those who were fortunate enough to have been screened, have suffered setbacks.

As I said, so many things have changed: more states are including expanded screening tests for newborns including that for GA1, treatment protocols are improving, and equipment and treatment for those injured are becoming more accessible. However, it does sadden me that even one child with this disease or the hundreds of other metabolic disorders will die or become injured because screening is not available to them. Like Nikki, they may appear perfectly normal on the outside.

I believe Nikki’s story is a small beacon of hope to families who have received the crushing news that their child may have a life-threatening illness. Over the years, I have been contacted by families from England, Alaska, and California to name a few, who have read our story and gained a bit of hope: so many of the stories out there have tragic endings. I have also received emails from teenagers who have happened upon her story on the internet and have ended up doing research or community outreach projects for school on newborn screening and from one girl who just happened to be looking up hits on Google on her own name and was touched to learn of our "miracle child." Nikki has been attracted to angels since she was very little. I am convinced that she has had several guardian angels throughout her life. Keeping Nikki healthy was our full time commitment from the day she was born. Looking at this beautiful 10 year old, we know that every sacrifice has been worth it.

My hope is that in the next decade, every child will be screened and more can have the life that Nikki has been fortunate enough to lead.

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Martha Duffy, Age 29, Propionic Acidemia shares her pregnancy and birth experience

Two years ago, I wrote an article for this newsletter detailing my experience with PA. At that time, I indicated we had just sent fibroblasts for mutation testing in an effort to help determine if my husband, Tim, was a carrier for PA as we were making decisions regarding having children. Thankfully, after almost a year, the test results came back showing that he was not a carrier. Thus began a journey that resulted in the birth of a beautiful baby girl, Grace.

My pregnancy brought with it both the expected as well as the unexpected. Although I have enough specifics to fill most of this newsletter, for time’s sake as I have a little one lying on me (who will want to be fed soon), a (relatively) brief summary of my experience follows.

Like many pregnant women, I experienced morning sickness throughout the first trimester. However, unfortunately, this didn’t end then but rather continued for the rest of my pregnancy. In addition to the usual remedies for combating morning sickness, I was given Zofran ODT. The orally dissolving tablets acted immediately to counteract the nausea and prevent vomiting as the potential for metabolic crisis would increase with continued, frequent vomiting.

I knew all aspects of the pregnancy would be closely monitored but I was surprised, both pleasantly and unpleasantly at what this brought. Although I did everything I was supposed to, I was still impressed at how good my metabolic labs really were; I don’t think they have ever been that good. Granted, at times results were low or high necessitating a change in therapy. Most of these changes were expected as the pregnancy progressed to provide enough nutrition for Grace. An ultrasound at 18 weeks showed bilateral choroid plexus cysts and suspicion for a VSD (ventricular septal defect) which, if present, increased the risk the fetus had a chromosomal abnormality. At this time I opted for an amniocentesis to determine if such an abnormality was present. Since an amniocentesis was being performed, we chose to have extra amniotic fluid collected and sent off to have PA status of the fetus determined. Thankfully, all results from the amniocentesis came back normal. Four weeks later, another ultrasound elicited concern for IUGR (intrauterine growth retardation). Although on paper I should have been getting adequate calories, I increased intake nonetheless. After all, it couldn’t hurt…only help.

I expected the toughest part of the pregnancy for me would be to eat enough calories and protein for adequate growth. What I didn’t expect was just how tough it really was. At any time throughout the day, I knew how much protein I had consumed and how much was left. The need to eat more than usual and on a consistent basis was extremely difficult and only complicated by my nausea. To meet increasing needs of the fetus without consuming more than my body could handle, I began taking a medical formula as well at the end of the first trimester. The amount needed was increased throughout the pregnancy. As my mild condition has not necessitated the use of such a formula when in a non-pregnant state, starting this, while nauseous, was perhaps the most challenging aspect for me. Needless to say, the taste left more to be desired. I am happy now to look in our refrigerator and, when I see the formula container, know that it is Grace’s formula to take and not mine. If any formula company representatives are reading this, I would be glad to provide some input for suggested improvements! I am hoping some changes in regard to medical protein options available by the time we are ready to do this a second time.

Given the risks involved, plans for delivery were formulated early on. With the efforts of those involved, I expected a well-controlled delivery likely earlier than 40 weeks. However, I did not expect delivery to occur at 31 weeks. This followed admission to the hospital at 30 weeks with preeclampsia which significantly worsened over a week and resulted in a c-section. Despite the fact that it occurred earlier than planned, the careful planning and attentiveness to detail of those involved resulted in a delivery that was well-controlled.

At 1:03 AM May 24, Grace was born. Weighing only 2 lbs, 9 oz and 14 inches long she was immediately taken to the NICU (neonatal intensive care unit) while I was taken to the ICU for post-partum monitoring. Eighteen hours after she was born, I finally got the chance to meet my daughter. Because of medicine given upon my admission to speed up maturation of Grace’s lungs, she did not require any respiratory assistance after being born. (And, as anyone who has been around her when she is hungry can attest, her lungs work more than well!) She remained in NICU for 7 weeks, primarily for feeding and growing. Weighing over 7 lbs, looking at her now it is hard to believe she was a preemie.

The success of this pregnancy was in large part due to the exceptional care I received. The importance of a good perinatologist (high-risk OB) and metabolic team cannot be underestimated. In my case, I was fortunate enough to have the best in both specialties. To Sally Segel, perinatologist at Oregon Health & Science University and to Jon Wolff, Sandy van Calcar and the rest of the metabolic clinic at the University of Wisconsin Waisman Center – thank you for all you did to ensure the best outcome of life’s most precious gift. Yes I had to eat more than I ever wanted and work through nausea every day but I couldn’t have done it without you. I look at Grace everyday and think about how fortunate she and I were to have your expertise to guide us through this journey. We are truly blessed in many ways; you are just one of the many. We pray she will continue to thrive and look forward to celebrating both life’s big and small milestones with her.

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Melissa Stagni, Age 18
Propionic Acidemia

Our daughter, Melissa Leigh Stagni reached a milestone this year – she graduated from Wayzata High School on June 5, 2007. This is a remarkable feat for a kid whose ammonia was near 2,000 when she was in a coma at 4 days of age! It is a miracle when we think of the many times we pushed her to the hospital to combat metabolic decompensation brought on by something as simple as a common cold.

Time flies by … the last update published on Melissa was when she was 11 years old, entering the 5th grade. This update will try and cover the last seven busy years!

Until the 5th grade, Melissa was fed through her NG tube during the night while sleeping. Her desire to spend the night at friends’ got us to gradually wean her off night feeds to a daytime schedule. She now receives four bolus of her special formula each day. Her formula still consists of Propimix-2, Prophee, 2% milk, water, 2 tubs of yogurt (18 grams of protein) and 1 jar of fruit baby food…mixed with water to make 1800 cc of formula each day. Melissa has been administering her own NG tube since was little and continues to do this four times per day. She has had no medical issues using her NG tube, so we leave well enough alone. When she was 16 we starting using the new Vitaflò Express PA/MMA product. She did not want to do her bolus at school any longer because she felt she was missing out on social time with her friends when she had to do to the nurse’s office to do her bolus feeds. She drinks the Vitaflò PA/MMA formula during lunch and uses the Tropical Punch flavoring. She likes that her friends participate and ‘shake up’ the cup for her. For those who aren’t familiar with Vitaflò, it comes in small packages and is an amino-restricted formula with about 18 grams of protein, but little calories. Melissa still requires high calories in her daily diet, so she still will take three bolus of her formula in addition to the Vitaflò. During the past 2-3 years Melissa has also had an enormous increase in her oral intake of solid foods. She is eating popcorn shrimp (not on a daily basis), mashed potatoes, pizza, hot dogs….just about anything - she's trying all sorts of new things. She took several cooking classes in school, which has over 3,000 students!

As you can see from looking at her graduation picture, she has earned her ‘letter’ from the high school athletic department. We are quite fortunate because Minnesota is the only state in the union that has an ‘adapted athletics’ sports program. This program is divided into two leagues – PI (physically impaired) and CI (cognitively impaired), for students with an IQ below 70. Melissa qualified for either, and chose to participate in the more athletically challenging CI program. Melissa played soccer, floor hockey, softball and bowling. She was the captain of her team in each sport. She was the starting pitcher on the softball team and had many strike-outs to her credit. And she was part of the bowling team that won the State Championship her Junior year. Through her participation in these varsity sports, Melissa learned valuable skills in good sportsmanship and teamwork. She also became widely recognized in the school, which has over 3,000 students!

In addition to her high school sports, Melissa has been a regular participant in Special Olympics since she was 8 years old. She participates in gymnastics, golf, basketball and bowling…and has earned numerous medals that reflect her dedication and hard work!

As I am sure you can tell we are very proud of the achievements that Melissa has gained over these years. She is a leader and role model among her peers. She has overcome many obstacles put before her as a result of her Propionic Acidemia and has become a great young woman. A woman that we admire and love very much. I would like to acknowledge and thank her metabolic physician, Dr. Susan Berry and dietician, Dorthey Markowitz at the University of Minnesota. Dorthey has been Melissa’s dietician for the past 18 years! We really could not got Melissa to this point without the two of you and we appreciate everything you have done for Melissa (and us)!

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Renewal Time!
Thank you for the many years of support you have given to the Organic Acidemia Association. We ask that you take a moment to review your membership today which will help continue our valuable work providing information and support for those afflicted with organic acidemia disorders.

Suggestion donations are $25 for domestic addresses and $35 for International addresses.

Summer 2008
We have found a major sponsor for our next family conference! Thanks to Dr. Jerry Vockley and Lynne Wolfe for assisting us in sponsoring our next OAA/FOD Family Conference which will be held next summer in Pittsburgh, PA. Details are in the works and we are happy to join the FOD Family Support Group again for our next conference. More information will follow in the next OAA Newsletter. If you would like to volunteer to help - please let me know!

Fundraising Bracelets
Brand New! Organic Acidemia Association fundraising bracelets will be available soon. Our previous fundraising bracelet was a combination OAA/FOD bracelet. This year we have purchased bracelets solely for the OAA – check the OAA website soon for more information and ordering procedures!

Newsletter Editor
If you are interested in helping assemble the OAA Newsletter - we need your help. Please contact Kathy Stagni at oaanews@aol.com if interested.