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Organic Acidemia Association Newsletter

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SAVE THE DATE
July 30 & 31, 2010
Atlanta OAA Conference

Thanks to the generosity of Emory University School of Medicine and our premier sponsor Vitaflo; plus Nutricia, Ross/Abbott, Sigma Tau, Rare Disease Therapeutics, Cambrooke Foods, Med-Diet, and other sponsors and exhibitors; we will be holding our next parent conference in Atlanta, GA on July 30 and 31, 2010.

We will be holding the conference in conjunction with the FOD (Fatty Acid Oxidation Disorders) Family Support Group. Info is posted on our websites (http.fodsupport.org & http.oaanews.org) but already we have a great line-up of metabolic nutritionists, doctors, other specialists, plus cooking, low protein food, and formula demonstrations.

The conference will be at the Emory University Conference Center Hotel and we have also arranged a discounted room rate at the hotel ($99 per night). You will need to make your own hotel and travel reservations as soon as possible.

Please see registration form inside & mail your registration before June 15. See you there! We'll have a **'Peach of a Time in Atlanta.'**

Spring Greetings from Carol

As always, time has flown by since the last newsletter. In February Kathy, Deb, and I attended a NBS Connect planning meeting in Atlanta. This is an exciting program being created to connect parents with researchers and we'll keep you informed as this program develops. In just a week from the time I'm writing this, and probably by the time you read this, my husband and I will have represented OAA and FOD at the annual American College of Medical Geneticists convention and the Society for Inherited Metabolic Disorders meeting, both in Albuquerque, New Mexico. Also, in April an OAA parent will represent the OAA and FOD at the Genetic Metabolic Dieticians International meeting in Baltimore, Maryland. We will report on all three conventions in the next newsletter (which should be published at the beginning of August).

These conventions are not only a way to learn more about rare metabolic diseases, but they are also a great way to get the word out that we are available to provide moral support to newly diagnosed parents. The comment I hate most to hear is from a parent of a 2 year old, who says she didn't realize we existed and that she really wishes she'd known about us earlier. I know how lonely I felt when my own daughter was diagnosed in the days before the internet, and want to help others not feel so alone. Note that this marks volume XXVII of the newsletter. That's 27 years of service! A lot has changed over the years but you can still view a decade of newsletters on our website.

Meanwhile, check above and inside for information on our upcoming conference. For those of you who can't attend, we will be posting speaker presentations on the OAA and FOD websites, and summaries will be published in the December newsletter. I must say that I have been impressed at how receptive the professional community of doctors, therapists, and nutritionists have been for our conference. We had many people contact us to volunteer to be speakers and to simply attend the conference with you, the parents. I have also been impressed with the sponsors. Even in these difficult times they have come forth with donations. Thanks to their generosity we have been able to keep the registration cost at what it has been for the last decade.
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Piper Clayre Pinkett

CblC, 7 years old

Piper was born July 16, 2002 after an uneventful pregnancy, except for my craving peanut butter and bean burritos. Actually, her birth wasn't exactly flawless. She was a scheduled c-section for July 15 and the hospital sent us home because they were not able to deliver any more babies that day due to the nursery being too full. Have you ever heard of such a thing? Anyway, she was delivered, Apgar scores were good, and all was going along very well until about 8 hours after birth. Piper was acting very sleepy and wouldn't nurse very often. This first night was high-lighted with two feedings and lots of sleep…for Piper. The next day, the lactation nurse came to visit due to my concerns and tried to reassure me that it would get better. She stated that Piper latched on well and all was going to be okay. We went home, but I was worried about her sleeping so much.

Piper continued to sleep more and eat less. Additionally, she developed some reflux after eating. Not projectile, but sort of oozing. It was time for her 10 day checkup, so I was relieved to get her in to see the pediatrician. When the doctor told me that Piper had lost 15% of her birth weight, I was astounded. Then he started asking me lots of questions about her feedings and stated that she was "failure to thrive." I began crying. Based on my experience, failure to thrive was for babies whose parents did not attend to them appropriately. I was forcing a bottle into Piper's mouth every three hours just trying to get her to eat… she was "attended to" quite well! The poor doctor tried to reassure me and ordered a series of lab tests. After determining that she had a UTI, she was admitted to the hospital for a few days and placed on IV antibiotics. Additionally, she was placed on infant formula, instead of breast milk, and sent home with another round of oral antibiotics. Reflux meds were also added to her daily regimen. Piper was sleeping less and eating more, so the doctor attributed her difficulties to the infection.

Feeding and sleeping approached a more normal routine and the reflux was better. After the sixth to seventh day of the ten day antibiotic prescription, I noticed Piper was sleeping more and eating less in quantity and frequency again. I had been letting her sleep on my chest each night since shortly after birth so that I could quickly offer her nutrition. By about the fifteenth day out, I was feeding her with an eyedropper because she no longer sucked and putting cool compresses on her to wake her to eat. Nothing was working. She was now foaming at the mouth with her formula, rather than throwing up. After a brief discussion with my mom, I decided not to wait until the following day to take her to a scheduled appointment with the pediatrician. My sis-in-law and I drove her to Arkansas Children's Hospital ER.

Immediately when the nurse saw Piper, I knew something was terribly wrong just by looking at his expressions. Her temperature was very low, she had turned light gray in color, and she never roused to any stimulation. Piper was taken to a room within seconds. The staff began asking lots of questions and trying to obtain blood. It took fourteen sticks to get an IV placed…Piper never stirred. In all the flurry of people, someone mentioned "unconscious and severe dehydration". Spinal fluid was finally obtained after eight tries. Her tiny little body didn't move on the gurney the entire time.

After a very long and stressful night, Piper was admitted to Arkansas Children's Hospital the next morning. Most of the next two weeks were a blur…lab work, new formula, tests, x-

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Piper Clayre Pinkett (continued from previous page)
rays, more new formula, and an MRI. The MRI came back as "odd", she was found to have a subluxed left hip, and she required a Ladd's procedure to correct a slightly twisted intestine. While in surgery, she had a g-tube placed. Post-op was fine and she was moved back to her room. Within 36 hours she was VERY sick and was taken to PICU.

The doctors began trying to find "zebras", as they called it. A zebra was the term used to refer to rare diseases, disorders, conditions, etc. Bone marrow and spinal fluid samples were taken. She came back with an unexplained purple crystal in her marrow, but this was not their major concern. Piper, at about the age of 35 days, was deteriorating daily. Eventually, a very nice doctor came by to get a detailed family history. She stated that they now had ideas that something genetic was going on. Piper was on TPN due to the new g-tube placement and now required mechanical ventilation. Later the next week, the other geneticist, Dr. James Gibson, came into our lives. He asked even more questions about our family history and Piper's symptoms. He smelled, examined, and scrutinized Piper very closely, and ordered more tests. He stated that he had strong suspicions about what was wrong with her but wanted to wait for the test results. Within a day, he came back and delivered the news. Piper had all the signs of Methylmalonic Acidemia with Homocystinuria. He said she would begin treatment as if diagnosis were certain, explained that not all patients were responsive to treatment, and asked permission to do a skin biopsy from her upper thigh. He stated this would be sent to a lab in Canada for confirmation.

Well, as they say, the rest is history. Piper began the Betaine, Carnitor, Leucovorin, Hydroxycobalamin IM regimen so common to children with Cobalamin C. She was introduced to a special mixture of medical formulas containing Propimex, Neocate, and SHS Amino Acids. She was off the ventilator within three days.

Her daily medication list has grown as much as she has. Due to her compromised immune system (not common for CblC), she takes Zithromax almost year round. Additionally, she takes Keppra daily for seizures. Singular, Zyrtec, Albuterol, and Rhinocort Aqua are for allergies/asthma, as needed. Hydroxyzine is administered near bedtime, along with Melatonin, to assist her with sleeping. Recently, choline, magnesium, and fish oil supplementation were added.

Piper hasn't been underweight for at least five years now. She weighs in at around 56 lbs! She is completely dependent on tube feeding at four hour intervals except at night when she gets a very slow feeding via pump. Piper is a professional taste tester, although when she ate by mouth, she was protein restricted. Her favorite flavors are those that are salty and dark chocolate.

As far as her development, due in part to a severe hypoxic event at around the age of 14 months, Piper is extremely delayed. Each week, she receives four hours homebound special education services and 3x45 minutes each of home-based OT, PT, and speech through our school district. She uses a wheel chair and is non-verbal, has scoliosis, and a dislocated left hip. Despite ALL of her issues, she is a social butterfly. She would rather play and "talk" to a person than a toy or object. She babbles a little, laughs and smiles most of the time, and stomps and postures her feet when she is getting aggravated. Additionally, she is quite the manipulator. If posturing and vocalizing don't work, she just lowers her head and pretends to be asleep. It's quite comical…though not to her therapists.

Overall, despite all the close calls and vertical learning curve regarding the field of medicine, Piper, who is now 7 years old, has been one of the best things to occur to our family. With the exception of injections, her brother loves her dearly and can care for her every need, including diapers! Every family member, immediate and extended, has become a bit OCD about illness exposure and hand washing, especially when they may come into contact with Piper. We've been known to live like hermits to avoid germs and sickness during certain times of the year. Additionally, they are all fabulous in times of need. Whether it is bringing me a change of clothes and tooth brush, providing a period of respite for me, or keeping me company as we sit beside Piper in the PICU, we are so fortunate to have such a great support system.

As many families of medically fragile children know, it takes a village to raise our children. If not for the patience of a few extremely dedicated, caring, and brilliant medical professionals, Piper would likely have left us a long time ago. When I've asked the same question three times, posed three different ways, Dr. Lucas, Dr. Gibson, and Dr. Kahler have never appeared irritated. The genetics specialty nurse, Paula, is instrumental in coordinating Piper's many follow-up appointments. Her nutritionists have gone down to the hospital nutrition room to ensure that her formula was from a fresh can, rather than an expired one. A particular phlebotomist, Cynthia, is sheer perfection in obtaining what seems like vats of blood from just one stick. Our local pharmacy goes above and beyond making sure her medicines are kept in stock, even having LONG conversations with suppliers who didn't deliver as promised. Finally, Arkansas Children's Hospital and Arkan-sas Pediatrics of Conway always place Piper's individual needs high on their priority list when she is visiting or inpatient. To these, and many more, I wish to extend a huge "Thank You!"

Finally, I wanted to say that I appreciate all that OAA offers. It was the very first site that my brother found after Piper was diagnosed. Although not always a rosy picture, I've appreciate the diversity of information and availability to link up with other parents. Keep up the good work and thanks for letting me share!

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Kumiko Elizabeth Ariella Menopi Boscarino –
MMA Mut 0, age 8

Kumiko was born September 12, 2001. I was 41 years old so we had an amniocentesis done. Without any history to indicate a MMA they didn't think to look for such a rare disorder. My mother is Japanese & my father is of English descent. Kumiko's father is a mix of Irish, English, Italian, & American Indian.

Kumiko's two half-sisters were born when I was 19 & 21 years old, so any additional discomfort I felt during pregnancy, I attributed to my age. This pregnancy did feel different & I found that I couldn't eat meat from around the 5th month on. I breast fed her in the hospital, but when we got home she refused to eat. We took her to the doctor & she took a bottle for him so he told us to give breast milk in a bottle. During the next few weeks she was still not growing well, so the doctor recommended adding formula to the breast milk.

My husband was very happy with our new baby & thought the doctors were being unfair in thinking she should grow faster. I was very concerned that Kumiko didn't look at our faces at all. The doctor was concerned that she was very floppy. By December, we'd seen the doctors quite a bit & she was still below weight, still not sitting up or holding her head up, & she seemed to spit up more than other babies. We thought every babys different but she just didn't look happy.

One Friday at 4 mos old, Kumiko became reluctant to eat & threw up a great deal of each feeding. By Saturday we talked to the nurse who thought she might have some sort of flu. We gave her water & Pedialyte. She got to where she couldn't hold down formula. She didn't have a fever, actually, she felt a bit cold to the touch. Sunday night, her breathing starting to get faster & faster & we decided to bring her to the ER.

Luckily, the hospital closest to us was Denver's Children's Hospital. As soon as we got there they whisked us into a room & gave her oxygen. They suspected elevated ammonia in her blood & proved it with a blood test. They put her on a glucose drip with added bicarbonate to adjust her pH. They told us they suspected a metabolic condition. Kumiko looked terrible, but the monitors showed that they were getting her stabilized. By noon she had several IVs & was sleeping though everything.

They put her in ICU with a dedicated nurse monitoring her. I was so tired that I went home, & my husband stayed behind. We took turns working, sleeping, & staying with Kumiko. Within a few days they had a diagnosis (MMA mut 0) & had a special metabolic formula for her. Kumiko woke up & her condition stabilized even more, to where we felt as though we were meeting our daughter for the first time. One medicine that continued & would become daily was L-carnitine, which helps remove the Methylmalonic acid from her blood.

The doctors suggested that we have a gastrointestinal feeding tube or "G-tube" installed so that in the future we could keep her hydrated even if she refused to eat. After nine days in the hospital we finally got to take Kumiko home. She was feeling some pain from the surgery, but as she started to heal, she started to show more signs of alertness than she ever had before. She was like a new baby. She began to look at things with interest & slowly she began holding her head up.

After discharge there were weekly blood tests, & many doctor visits & prescript-ions. One of Kumiko's prescriptions was for B12 injections. Not only was this a weekly shot that hurt, but the first pharmacy gave me a cyano type, so the doctor made me get another kind from a special pharmacy. In the end she turned out to have a type of MMA (Mut 0) that doesn't respond to B12. The pediatrician recommended something called Emla or Lidocaine to reduce the pain of the shots. The Emla cream goes on the surface of the skin about 20 minutes before the shot, & then you cover it with a Telfa bandage to keep it from rubbing off (I used plastic wrap because it's cheaper). Now, when I pull out plastic wrap Kumiko knows shes about to get a shot & starts screaming. The most important prescription was for the low-protein formula: Propimex from Ross. (Later we used XMTVI & DuoCal.) We were lucky the State of Colorado passed a law requiring our insurance to cover special formula.

Another big adjustment was the g-tube. My husband & I both took the 20 minute class, but neither of us felt prepared. In theory, we were supposed to feed her by tying a syringe above her crib & letting formula flow into her by gravity. In practice we found that it was impossible to keep her still & we ended up with more formula in her bed than in her. It was more practical to push the formula into her using a syringe, but you have to watch her face for signs of gagging as you go.

Another thing we learned about the g-tube equipment is that they don't give you enough, so you have to learn to clean the tubes out & re-use them. We set up a hook over the sink for drying the washed tubes. We found that we could extend the life of a syringe by putting a little canola oil on the plunger after washing it. Also, lucky for us, Denver has a lot of stores for people who make home brewed beer, so we were able to find tiny, good quality pipe cleaners there. We learned to always keep at least one brand new syringe in her diaper bag in case one broke & two wash clothes for catching the drips.

We were told that the g-tube sometimes leaks & we were given a box of gauze & tape each month. We found that the gauze seemed to hold the moisture near her & make it more irritated. It seemed to work better to just towel her clean each time we fed her. However, at around 18 months old, we started having more & more trouble keeping the area clean enough. She had an ongoing rash that we were treating with lots of gauze & diaper cream. Then one day, the part of the g-tube that closes the hole broke off. I made a temporary plug by taking a plastic tube tip, heating it & pressing the end closed. The new plug worked better than the old one & her rash cleared up. (cont'd next page)

Kumiko (continued from previous page)

Kumiko has never learned to feed herself. She did eat by mouth for a few months & we only used the g-tube to feed at night. She started eating solid foods at around 6 months old but after a few months, she started refusing to eat anything by mouth & I sometimes wonder if things would be different if she'd been getting hungry between feedings like other children. In any case, now that she has a g-tube she can only be watched by someone who knows how to feed her. The school she attends must have a full time nurse on staff. Also, Social Security has programs that help pay for daycare but because of her g-tube they require that the care be administered by an LVN or better. With the nursing shortage, it's impossible to find an LVN who will accept the sitter pay, so Kumiko doesn't get the service.

For several years Kumiko did pretty well, she got periodic blood tests to monitor her amino acid levels, & was hospitalized a couple times a year. In between she was a pretty happy child, if somewhat weak & behind in her development. We got a wheel chair because she tired easily walking long distances (like through a mall, although walking there is one of her favorite things to do). She also loves swings & roller coasters.

In December 2005 she had a mild flu, & we brought her to the hospital as a precaution. The next day after tolerating her formula, & tests showing no elevated MMA or ammonia, she was released. But she wasn't better. 36 hours later, Kumiko was lethargic, vomiting, dehydrated, & definitely on her way to a crisis. Although we keep a copy of Kumiko's protocol letter in her "hospital bag", we forgot to check to make sure it was still there. Instead, it was with her discharge papers from the previous day & there was a delay. She went into a full blown crisis & was hospitalized almost 2 weeks.

During her stay they administered an antibiotic that caused a rash all over her body and really drew out her stay. The doctor put her on insulin because she was getting dextrose via IV, then had the nurses check her blood sugar every 15 minutes. This went on for days & she couldn't get any sleep & felt terrorized by it. After a few days I learned that the nurses could skip the toe prick if I had them write "parent refused" in the chart. It made the doctor mad, but it allowed Kumiko to sleep.

Although she had been able to walk & was working on stairs before going into the hospital, when she got out she couldn't even stand for weeks. A nurse told me to expect a week of recovery for every two days in the hospital & that's about what it took. She looked so sad for so long I worried that she might never be happy again. She also stopped talking for weeks.

After that, Kumiko went about two years doing pretty well, back on her old routine of hospitalizations occurring about every nine months & lasting two or three days each with no serious complication. The state sent us occupational therapists who helped get her playing, moving, & interested in new things. By then we were living in Los Angeles where they have excellent special ed schools & other things for special kids.

Then in 2007, her father, who had been her primary caretaker while mom worked, graduated school & started working as a junior high school teacher. By that time the three of us had moved in with other family members. There were people around to help take care of Kumiko but our communication was a little lacking in the area of diaper changes – no one noticed that she had stopped pooping. She started having bad constipation & making sure she got enough water started to become a real problem. The doctor also started making big changes to the volume & protein level of her formula. It took working with the doctors & really focusing on water & making sure to give her lots of laxative, before we were able to get it under control.

Now Kumiko is 8 years old. This year for the first time, she is in a special ed classroom in a general education school, & has a tough teacher who makes her work at learning to read, write & catch up with her grade level. She is unable to dress or feed herself, & only recently & somewhat rarely engages in "mean-ingful speech", although she sings all day long. She loves water, & will pour it on her head anytime she can. She also loves salted popcorn which she doesn't eat but just sucks & discards. She loves picking flowers at the park & "climb-ing trees" (really I just put her in a tree & let her enjoy the view), & she never tires of tickling.

Last year, Make-A-Wish granted Kumiko & her dad a week's vacation in Orlando where they spoiled her & lavished her with gifts. We also went camping with her older sister & to a water park for her birthday. She loves playing with her computer, watching her favorite videos, goofing off in the yard, & spending time with Mom & Dad. All in all Kumiko is a very happy kid. If you're looking at a child in the hospital right now you may find it difficult to imagine seeing that person smiling & playing with abandon someday, but I felt that way too. Just give it time.

Kumiko's dad & I fight hard to minimize the doctor visits & blood tests in her life, hoping to maximize the vacations, park visits & everyday fun. I don't take the things doctors tell me as gospel, I believe that you must critically assess everything they say & judge for yourself what priorities to hold dear. You & your family have to decide how to fit these challenges into your life & what to move aside for it. But every family is different.

We just wanted to end our story by saying that when Kumiko was first diagnosed, it was frightening. One of the scariest things was how little information was available. I don't mean technical information like how the disease works, but simpler questions like what kind of life she might have, & how we could help her. When we found the OAA & read what other parents had written about their children, their progress, & what their lives were like, it was a great relief. So, now it is my turn to try & help others with our story. If you have a child recently diagnosed with MMA or similar condition, believe me when I tell you that while everything might seem overwhelming at first & there is always some stress, soon it will become easier.

Every life has challenges & when they don't, those people usually go out & make some for themselves. Your challenges will be interesting & the rewards will be great. Good luck!

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Cady Pierce (Propionic Acidemia, age 13)

Oh, where to begin.. I can't even remember the last time I wrote about Cady. Although, I've got a lot to write about! In July of 2008, we went on vacation to Ocean Isle, NC. While there, Cady became constipated. We did what we could to clean her out, but this was no ordinary bout. Cady generally doesn't have problems with her ammonia levels until the end of a spiral, when all else is lost and there's no turning her around without major medical intervention, but this caused her ammonia to spike. She continued to be constipated throughout the summer and fall, with alternating bouts of high ammonia levels, constipation and then higher and higher doses of laxatives. It was a vicious cycle and I did everything I could to get it under control. It came out of the blue, without change to her diet or activities. I saw her pediatrician, then on to the geneticist, then a consult to the University's GI clinic. All anyone would tell me is "give her more laxatives." I could never get anyone to understand this wasn't like Cady, there had to be a *reason* for the constipation, that we need to get to the bottom of it instead of giving her more and more laxatives. Right after Christmas, I had a major abdominal surgery. Of course, that is exactly the time that all else breaks loose too. Cady got the mother of all constipations, and again the advice was "give her more laxatives." I stressed that I was giving a 60 pound child enough laxatives to keep the nursing home flowing freely into next year, but no one had any answers. Cady "blew out" and dumped all of her electrolytes in the process. It was more than her embattled system could stand, and she spiraled on us big time. We called an ambulance, she was barely conscious and slipping further and further away. There was no way to get a gurney into our house to fetch her, so without any thought to myself at all, when I saw the lights pull into our driveway, I threw all 60 pounds of Cady over my shoulder, ran down the front steps with her, and climbed into the ambulance. It didn't occur to me until later that I still was on an 8 pound lifting restriction.

In the ambulance, Cady's blood sugar was tested out over 500. She normally doesn't have a blood sugar problem, and some IV fluids were begun. By the time we got to the ER, just a few minutes away, her blood sugar had come down to the 300's. She became a little more awake, although she was confused and tried repeatedly to pull her oxygen mask off. A decision was made by the local ER to fly her to Morgantown. In all the times she'd been sick, she'd missed the bird. Now it finally got her. In 20 minutes she'd be in Morgantown, and I was off to pack a bag so I could stay with her. It was a 2 hour deer-dodging drive for me in the middle of the freezing night. By the time I got to Morgantown, she was in full coma, and it would be a week before I'd hear her voice again.

While the PICU in Morgantown was a Godsend, (they had built new suites with captains beds for the parents to stay with their child), much of that week is a blur to me. I am not one to shrug things because they are painful, but I cannot go back to those memories without tears. Cady lay so helpless in that big bed, its siderails padded because her sodium was so low that she was apt to have seizures at any moment. Her foley bag filled up again and again, despite only having a little IV fluids running. She would not wake, although she would moan to very painful stimuli. Once again, specialist after specialist was called in to evaluate this perplexing child. Her acidemia had all but resolved, and yet she remained in coma. There had been some discussion that maybe there was cerebral swelling causing her to dump all of her fluid, so she was CAT scanned. By the next day, the consensus was that while her brain was swelled, the problem was actually SIADH. SIADH is syndrome of inappropriate antidiuretic hormone. It is a condition of self defense gone awry, and deadly. For whatever reason, the adrenal glands begin to flush all of the sodium and water from the body. When these shifts occur, the brain swells, leading to seizures, coma and death. It was like Cady was saying "You think PA is confusing?? Try this!". And she wasn't done…

The treatment for SIADH is careful electrolyte replacement while restricting fluids to a bare mininum. This is very hard on veins, so a central line was introduced. While giving her pain medication so the central line could be placed, Cady stopped breathing. Her sedation had to be reversed and she began breathing again. The femoral line was placed and now we had a safer administration route for the concentrated electrolyte solutions. It became days of what I call 'chasing the monkey'. She'd get a dose of calcium and blood would be drawn. Then her magnesium would be low, and would be chased with a magnesium bolus. More bloodwork, more out of whack labs, more chasing. And still she slept. And all I could do was pray and pace and pray some more. So that's what I did.

As Cady's acidemia resolved, we began introducing calories to her. TPN was begun to help try to replace some of those crazy electrolytes as well. She was still having labs drawn, but now instead of her electrolytes being the focus, we began to watch her platelets. She'd had a blood transfusion already, the PA tends to supress bone marrow activity, so Cady wasn't making her own red blood cells at the height of her illness.. but now all of her cells were slowly recovering except for her platelets. Every day and a half or two days, she'd reach a critical level with her platelets, and need a platelet transfusion. We'd gone through this cycle three times, and couldn't figure out why her other cells were recovering, but her platelets were being chewed up so quickly. More bloodwork was ordered, and a hematologist consulted.

Everyone that came to see Cady that week (and she had a parade of visitors) prayed with us for her. All along, without each of us speaking of this to each other, the recurring theme was "she will be restored." I believed it, and held fast that God would continue to be with Cady.

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Cady (continued from previous page)

DJ stayed with us that week too, sleeping nights with Jay in a motel room until we got a room at the Ronald McDonald House nearby. I wanted him to spend as much time with his sister as possible. If he lost her, I wanted him to know why.. and if he didn't , I wanted him to appreciate why. After a week, I was exhausted, Jay was exhausted, we were at our wits ends. All along we had an enormous outpouring of support from friends and family, all of which offered to take DJ if we'd just say the word.. So, Sunday, after a week and with no end in sight, I called DJ's Autism Mentor from school and asked her to take him. Cady was due for another CT scan, an MRI for which she'd have to be sedated and intubated, and a bone marrow biopsy. I didn't think I could deal with anything else that day. DJ was going to get hungry, he was going to get bored, and underlying all of this was his Autism. He'd been an absolute angel all week, but it was time for him to go and allow us to concentrate on his sister. And since consistency is very important to Autistics, I wanted him to go with the one person who could be with him 24/7 until we got back home.

Cady had her CT scan, which showed swelling gone. They didn't have to sedate her for the MRI, but did afterward for the bone marrow biopsy. She was given a dose of anethesia, and we ran six floors to the ICU while bagging her through an ET tube. As we approached the ICU with her, I could see the latest bag of platelets hanging on her IV pole, ready for infusion. Just as the bone marrow biopsy was being completed, a halt was given on the platelet transfusion. It turns out some of the labs ordered with the hematology consult had begun to come back.. and there was an answer to the platelet problem, after all. Cady had developed HIT, heparin induced thrombocytopenia. There had been heparin in every prepared solution Cady had been given and she'd developed an allergy to it. This allergy caused her to destroy her platelets, but platelet reinfusion is the last thing you want in HIT. It will cause the platelets to aggregate together and throw blood clots throughout the body, which result in heart attack, stroke and organ shut down. Her platelets were low enough to cause spontaneous bleeding (which she did enough of when they pulled the ET tube from her throat that it looked like Freddy Kruger had been visiting) but we could not replace them. We just had to stand by with her on the precipice and pray she'd not bleed to death before she made her own platelets again.

After cleaning her up (like I said, she was a bloody mess after the ET tube was pulled) and washing her hair, we sat her up and braided a prayer cloth into her hair that my parents had brought from their home church. We weren't getting any reasssuring answers from the medical tests that had been performed.. the MRI only showed abnormality, which we already knew. If your brain swells, it looks different as the swelling is going down, and there's no real way to tell if the damage is permanent or will recede the way tingles leave a slept-on arm after a bit. All of the results were the same; "abnormal but we can't tell for how long."

Later that evening, we were waiting on the neurologist to come see Cady about an EEG that had been run the night before. She was late coming to Cady's room after her clinical day, and hadn't read the EEG yet. She said she'd read it and get right back to us. Jay and I waited an hour or two, but were famished and hadn't left Cady's side all day. I had decided I'd had enough ambiguity to last me an entire day, one more report was not going to say anything life changing. I told Jay that the only thing that would make me feel better was to give Cady a hug. I'd resisted it all week, she had so many wires and tubes around her, it made getting near her nearly impossible.. but I could stand it no longer. Jay got behind me so he could hug us both and I lifted her in my arms. As I lifted her body up to get my arm around her back, her head fell back and she opened her eyes. In that moment there was clarity, and she uttered "Daddy".. we cried like babies, and she fell back asleep. At that moment I knew all of our prayers would be answered, and we were on our way out of the valley.

Jay and I ate a small dinner, went to the local WalMart and bought Cady a Valentines bear that held a heart that read "Princess". When we returned to her room, it'd been two hours and she was exactly as we'd left her. I told Jay to get on one side of the bed and help me roll her so she wouldn't get a bed sore. At the sound of my voice, Cady sat up and began asking two hours worth of coherent questions. She asked them so quickly, we didn't have a chance to answer them. She'd been mute for a week and was gonna make up for it! The nurses couldn't believe it, it was the kind of thing that can only be called a miracle. Jay and I were so happy we could barely contain ourselves. It was the first good night's sleep Jay'd had in a week, and I'd say it was mine too, but she kept me up all night babbling.. still, I can sleep later, right? What beats having your child back? Nothing.

The next morning, the doctors and medical students who'd been following her rounded. They were just as amazed to see her sitting up, watching tv and coloring. The doctor that had admitted her smiled and said "Gee Cady, I wish we knew where your on switch was." We stayed in the ICU the remainder of the day, briefly unhooking and walking the unit, and then were transferred to the regular Peds floor the next day. The first order of business was to get Cady showered. The gals in the Unit did a great job of keeping her clean, but you all know what PA smells like after a while.. whew! While in the shower, Cady began to lose her hair in great clumps. The ordeal was finally taking its toll on her outwardly.

Cady spent the next day and a half on the Peds floor, just being watched, simply because no one knew what else to do.. who wakes up from a coma like it was a nap? On Friday, 12 days after this whole ordeal began, we were on our way home.

The following couple of weeks were hard on Cady and those caring for her, though. While she "snapped" right out of her coma, she regressed some after coming home. She was cross and wanted to sleep all the time. She couldn't walk very well and was no longer continent. We reverted back to g-tube feeds because she just didn't have the stamina to eat. Again, we did what we know.. we called a prayer meeting and prayed for Cady again. You can never get too much of that, you know.

Cady stayed home from school another week while I worked with her, and I sent her back to school. I knew Cady was still in

(continued next page)

Cady (continued from previous page)

there, I knew if I pushed her, she could answer the questions I asked, but it had been a hard go-round for her, and she just didn't want to. Well, ultimately it didn't matter what she wanted, it mattered what was best for her. I sent her to school with her feeding pump, and called her teacher. I warned him that it was going to be tough with Cady. She'd regressed to the point that she didn't want to do anything. I told him to ignore the tears as best he could, that she was in there and I was determined to have her back the way she was before "the fall". He called me from school every day. There were big crocodile tears, he wasn't sure pushing was the right thing. He wasn't sure she was up to it.. but he hadn't seen what I'd seen, and I told him to stick with it. Fortunately, there were two wonderful aides in the class and they helped shoulder the burden enormously. This was a team of adults that loved this little girl and was willing to drag her back kicking and screaming (which she did) and I thank God every day for that. The torment was relatively short lived. Within a couple of weeks, Cady came around to her old self. And it wasn't necessarily gradual either. There was a little progress each day, but then one day she decided we weren't going to give up on her and fighting all four of us was too much work. The tears stopped, and the smiles began. She excelled in her work, she was happy and back among us.

And that where we are today, a year later. Cady is a vibrant, 3-meal-a-day-eating, schoolwork-doing, Hannah Montana-loving 13 year old. She did suffer some permanent hearing loss due to the acidosis or electrolyte swings, and now wears hearing aids full time. Most of the time you don't even notice them. However, not only has she not been sick since that episode last winter, but her health is actually the best it's been in her whole life. Her labs are great, she's jumping grade levels in her work at school. She played Upward Soccer last spring, enjoyed Spring Break and summer at the beach and walked all over Manhattan in July. This year was one big roller coaster ride that left us emotionally spent, exhausted, exhilarated and enormously thankful. Oh, so thankful.

Jay and Leslie Pierce
New Martinsville, WV
Parents of Cady, 13, PA and DJ, 10, Autism.

Find me on myspace (PierceRN)
and on Facebook (Leslie Pierce)

Children's National Medical Center

NEW DRUG STUDY FOR PATIENTS WITH HYPERAMMONEMIA

Researchers at Children's National Medical Center are looking for children and adults to participate in a NEW drug study for hyperammonemia, or elevated blood ammonia levels.

Eligible patients must be:

- Diagnosed with carbamyl phosphate synthetase I (CPSI) deficiency, N-acetylglutamate (NAGS) deficiency, Propionic Acidemia (PA), Methylmalonic Acidemia (MMA), or Ornithine Transcarbamylase deficiency (OTC)*
- 1 to 70 years of age
- Willing to travel to Washington D.C. for a 4 day-long study and tests (travel and lodging paid by Children's National Medical Center)

If you, your child, or someone you know has any of the above diagnoses, you or they may be eligible for this study. Participation is voluntary and may help provide insight into an additional treatment option for children and adults with these conditions who suffer from hyperammonemia.

For more information, contact:
Dr. Nicholas Ah Mew: nahmew@cnmc.org
(202) 476-6177
Children's Research Institute
Children's National Medical Center
Washington, DC 20010-2970

*Patients with OTC deficiency must be *female*

'Peach of a Time in Atlanta'

FOD/OAA Family Support Groups
National Metabolic Conference

July 30 – 31, 2010
Atlanta, Georgia

In order for us to serve you better, please fill out form COMPLETELY Submit online or Mail to Deb with your check (made out to FOD) By June 15, 2010 or pay online via PayPal ($2 extra charge)

2010 FOD & OAA METABOLIC CONFERENCE REGISTRATION FORM

Location: Emory Conference Center Hotel (ECCH)
1615 Clifton Road Atlanta, GA 30329

Tentative Agenda

- Friday, July 30 Metabolic Sessions (Each Group in own room, lunch at 12) 8 am - 5 pm
- Friday, July 30 Reception & snacks ~ Great time for Family Networking 6:00 pm – 9:00 pm

Saturday, July 31 Wear TSHIRT Lunch & Combined FOD/OAA Session 8 am – 5:00 pm
Continental Breakfast in the designated break area from 7-8 am for Registrants only,
Lunch both Friday & Saturday in the Center Dining Room will be from 12-1 pm for Registrants only
For Agenda & Speaker Updates (when confirmed) refer to: www.fodsupport.org or www.oaanews.org

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CONFERENCE REGISTRATION FEE: ___ Paid by Check ___ Paid online via PayPal ($2 extra charge)

• Registration Fees cover the cost of meals & a special FOD or OAA T-shirt (ADULT unisex shirts small to xxlarge) . Continental Breakfast & Lunch will be served on Friday & Saturday for all conference registrants. . Snacks will be available during session breaks & at Friday evening Reception. . Low Protein &/or Low Fat food available: Please indicate special food requests by June 15, so we can let the Chef know the quantity needed.
You must be registered for the conference to be served the two continental breakfasts & 2 lunches & snacks.

Number of people attending_____ at $50.00 per person over age 13
or number attending_____ at $30.00 if you have an FOD/OA, or kids over age 5, or sitters for kids
*** [To cover their fee, if paying via PayPal, the Registration fee is increased to $52 & $32]***

$_____ enclosed – Conference checks made out to 'FOD' & mail to Deb (see next page or pay online via PayPal). There will be an unsupervised children's activity room (you will need to provide your own supervision by a family member or sitter & if you'd like them to join us for meals and/or snacks all must register below). Mature Teens are encouraged to attend sessions – all must register.

Name of Family Registrants state SPECIFIC disorder (ie., MCAD, PA, Unclassified etc) , & indicate (**) if you have an FOD or OA; Tshirt sizes – adult S, M, L, XL,2XL) Professionals register name on page 2.

	Tshirt Size?		Tshirt Size?
Name:_____ Relationship:_____ List *Specific* FOD/OAA:_____		Name:_____ Relationship:_____ List *Specific* FOD/OAA:_____	
Name:_____ Relationship:_____ List *Specific* FOD/OAA:_____	Tshirt Size?	Name:_____ Relationship:_____ List *Specific* FOD/OAA:_____	Tshirt Size?
Name:_____ Relationship:_____ List *Specific* FOD/OAA:_____	Tshirt Size?	Name:_____ Relationship:_____ List *Specific* FOD/OAA:_____	Tshirt Size?

Families & Professionals:
___ # attending Friday (8 am to noon & 1 to 5 pm) FOD or OAA session (circle one)
___ # attending Friday Lunch (noon to 1)
___ # attending Friday night Reception (6 to 9 pm) at ECCH (registered children can attend)
___ # attending Saturday joint FOD/OAA Sessions (8 am to noon & 1 to 5 pm)
___ # attending Saturday Lunch (noon to 1)

ORDER EXTRA SHIRTS for other family members at $10.00 per shirt paid by *separate check* made out to FOD (for FOD shirts) or OAA (for OAA shirts) & mailed to Deb, along with this form by June 15th or $12.00 per shirt via Paypal All shirts are ADULT size.

Extra # of ____ small ____ med ____ lg ____ xlg ____ 2xlg ____ FOD shirt or ___OAA shirt

$_____Total Extra Shirt Amt Enclosed _____ Paid by Check _____ Paid online via PayPal ($2 extra)

FOD & OAA Emory Center HOTEL RESERVATIONS: 1.800.933.6679 or 404.712.6565
Families & Professionals must make own hotel reservations & travel arrangements

**Special room rate: $ 99.00 plus tax (single/double)
You must state that you are attending the FOD/OAA National Metabolic Conference

IMPORTANT NOTES: When you make your Hotel reservation, please indicate any special requests (ie refrigerator, microwave, accessible rooms, etc.) The Hotel will make every effort to accommodate these requests based upon availability. Hotel has elevators to all levels. Wired internet is available in guestrooms (fee applies). There are complementary computers & printers for guest use located on the lobby level. Complimentary wireless access is offered on the conference floors & main lobby level.

FAMILIES: CONTACT PERSON INFORMATION. Please provide all information in case there is a question regarding your registration:

Name_____Address_____

City_____ State_____ Zip_____ Phone # ( )_____

Email: _____

SPECIAL FOOD REQUIREMENTS (low protein, low fat, food allergies):_____

____ # of children that might use Activity Room Ages:_____ Name of your sitter (s) _____
Are your children registered? ___Yes ___No Are your sitters registered? ___ Yes ___No
(Children under 5 do not need to pay but please let us know about them. Everyone needs to be registered to get food)

PROFESSIONAL Registrants: Please provide us with the following information:

Name _____ Title _____

Institution _____ City, State _____

Phone # _____ Email _____

[You may go between the 2 meeting rooms – just be sure to mark which shirt you'd like- either FOD or OAA]

_____FOD or ____ OAA size: ____ Small ____ Medium ____ Large _____ Xlarge _____ 2Xlarge

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_____I cannot attend, but please accept this donation for the meeting.
_____Volunteer to help with Conference ___Yes ___No

SEE YOU THERE!

Carol Barton, Executive Director
Organic Acidemia Association
carolbarton@oaanews.org

Deb Lee Gould, MEd, Director
FOD Family Support Group
PO Box 54
Okemos, MI 48805-0054
517.381.1940
deb@fodsupport.org

••• Mail Registration Form & all Checks to Deb's address •••

DUE by June 15, 2010

Trish Campbell (MMA)

Trish died on October 23, 2009. She decided after a ten year battle with Dialyses, and suffering through kidney transplant rejection with the knowledge it would be at least 6 more years until she would be eligible to receive another transplant, that she could not fight any longer. Trish put herself on Hospice on October 13, 2009. We were so blessed that she came to us, her stepfather Steve and I, and told us what her plan was. We were able to walk with her to heaven's door and love her through this the last journey of her life.

Her grandmother, grandfather and sister were here with her also. Trish was at peace with her decision and she researched her options making an educated decision with her doctors knowledge. We have heard from many physicians that this happens more often than we even imagined. Trish's Neurologist explained that especially with young people, facing the prospect of long-term, lifetime spent on dialyses, many make the decision to let go.

I know that Trish's life was a series of miracles. She was not supposed to live, according to the medical profession at the time of her birth she was on borrowed time. I was assured several times in her first year that she would not make it through the night. And, yet, we had her for 36 yrs and 10 months exactly.

I am grateful for all the connections that Trish was able to make, especially within OAA. The support, she recieved was invaluable to her. I am attaching a photo, it is the one she wanted us to use on her memorial. Also I am attaching a poem her sister worote to memorialize her.

I miss my precious daughter immensly and there will always be an empty space in our live's, now she is gone. I thank the Lord that Trish found you, all. Trish was truly a blessing, and she taught me so much about life and acceptance of God's will.

Again, Thank you all, for all you meant to her and all you do for other families struggling to understand, the gift they have been given. I believe the Lord only gives us as much as he can handle and what we have to do is turn to him and trust that his will be done.

The problem us humans have is that we think God, only gives us as much as we can handle and we try to handle it all without him, or do like I did and get mad at him. I am grateful that God allowed our lives to be blessed by my precious daughter, and I am grateful she no longer suffers, but that she is finally singing with the choirs in heaven. No more pain, No more tears, only God.

My Sister, My Friend

by: Karen Perkins 10/25/09

She wasn't just my sister, she was in fact my friend.
We shared a special bond, right up to the end.
Feelings that weren't spoken were always understood.
No matter how bad the situation, we both could find the good.

Of course now we were sisters, so at times we disagreed.
We always could work through it, time was all that we would need.

Her strength matched her beauty, down to her very heart and soul.
She made the very best of life, never expecting to get old.
She found flavor where none was, she found beauty that escaped all the rest of us.

She always had a plan for each and every day,
and disliked it very much when it didn't work her way.
Never willing to settle, she always searched for more.
And I know that she found it on heaven's golden shore.

The pain that she did suffer has all been washed away.
I know that she rejoices, for peace she found that day.
Our family gone before her, they met her at the gate.
They were so glad to see her and let her know it was worth the wait.

My sister she lives on in the hearts of all she knew.
A little piece of me is now a piece of you.
You know that if she loved you that she loves you still.
The fact that she's not here with us,
Well, that is just God's will.

My sister she's an angel sitting at God's right hand.
She left to go and help carry out his perfect plan.

When the wind blows gently by you,
and the air smells a little sweet,
SMILE
My sister came and gently brushed your cheek.

Don't be sad she told us,
and please try to understand,
it was her time to go,
this was her final plan.

Hold on to your memories they will surely make you smile.
We know that we will see her, even if it takes a while.

So, dance when it is raining,
sing aloud when you choose,
make the best of what your given,
thats what my sister used to do.

An Education on Special Education

As the parent of a child with special needs, I can say that second to medical-related issues, ensuring that my child receives an appropriate education is one of the biggest and most time consuming battles I fight each year. It involves a tremendous effort to coordinate and collect all the required paperwork from medical specialists and provide copies to educational specialists and therapists, and vice versa. Furthermore, synchronizing schedules between all of the team members to meet and hold a conference is a daunting task. To add even more anxiety into the picture, keeping abreast of all of the special and general education rules and regulations is nearly impossible. Lastly, learning to compromise, prioritize, and pick my battles is the most difficult part of all. My saving grace is that I have some insider information… that is, I **AM** a special educator… specifically, a school psychologist.

In considering how to begin this article, trying to nail down a starting point to what seems to be a never ending and cyclical topic was my biggest challenge. With the motivation of wanting to assist parents like me in being able to navigate the special education maze without all the confusion and make more informed decisions, where was I to start? I have an unwavering belief that parents are the best advocates a child can have. That being said, there are parents who actually hinder their child's access to educational services, sometimes accidentally due to lack of knowledge and sometimes unintentionally because they just didn't have the right understanding of special education. So, it came to me that the special education process actually does have a beginning and it would likely be best if that is where we start.

Per the idea.ed.gov website, the "Individuals with Disabilities Education Act (IDEA) is a law ensuring services to children with disabilities throughout the nation. Infants and toddlers with disabilities (birth-2) and their families receive early intervention services under IDEA Part C. Children and youth (ages 3-21) receive special education and related services under IDEA Part B" of this law. If your child has disabilities that impair his/her development and/or ability to receive an education, your child may be entitled to special education and related services.

Here's a bit of parental input. Early on I learned that I needed to become BFF (best friends forever) with my email, digital voice recorder, and the copier. It's a matter of self-preservation for both me and my daughter. There is absolutely no way that I can remember 100% of who said what to whom and their response! It allows me to maintain a transcript AND filing system. Even my voice recordings can be downloaded to my computer and sent to my email account for filing. I keep everything in a "Piper" folder – an electronic one in my email and a paper one for correspondence and records that don't originate electronically. This works for everyone she comes into contact with: doctors, nurses, teachers, school administrators, therapists, and others. If they have email, I prefer to use it. Don't let me fool you. I really want to be organized…which I am not, BUT it sure looks that way to others when they see my three ring binder or are over my shoulder or on the phone when I need to send something to them electronically from my email. Perception is more important than reality…sometimes!

Well, now we get back to the topic of special education. It has been my experience that medical professionals, especially geneticists and genetic counselors/nurses, do a great job of informing you that your new baby/infant will likely benefit from early intervening services. So for now, I am going to just mention that if your child aged birth-2 may have difficulties, apparent or implied, seek assistance from various sources immediately. This should include at least a call to your state department of education and department of human services so that they may provide you with a list of early intervention service providers. Your child's pediatrician will likely know of reputable therapy providers, in addition to the fact it requires their prescriptions for assessment and therapy services to get the ball rolling for most insurance companies. At this point, services are usually clinical in nature. Furthermore, your child's school district (local educational agency or LEA) is responsible for ensuring that all children with disabilities from birth to 21 years within its jurisdiction and in need of special education and related services are identified, located and evaluated, regardless of the severity of their disability. This is called Child Find. I would hazard to guess that most states do this in a similar manner to ours by notifying parents through publications or announcements in newspapers or other media, with local circulation adequate to notify parents throughout the LEA's jurisdiction.

The Individuals with Disabilities Education Act requires that states, as a condition of eligibility to receive federal funds, provide special education and related services to eligible children with disabilities beginning at age three (3). When your child is aged 3-21, special education and therapy services (called related services) can be a bit trickier to understand and access. There are requirements that are specific to ages 3-5, 5-21, and 3-21. If your child had received early intervention services and had an IFSP (individualized family service plan) prior to age three, then an evaluation and determination of eligibility for services will likely transpire for the 3-5 aged bracket. This occurs when the child transitions from the preschool setting to the public school setting for kindergarten, also. Most refer to this process as referral.

A referral is warranted when a child is suspected of having a disability that adversely affects his/her education, and special education and related services are considered necessary to meet the child's individual **EDUCATIONAL** needs. The referral may be requested by a parent, a teacher, or some other person who is knowledgeable about the child. It must be in writing and requires a conference in order to make a determination regarding the child's necessity for an evaluation, no evaluation, and/or temporary placement in special education services due to unusual circumstances. One of the most difficult aspects of transitioning from early intervening services (Part C) to the 3-21 (Part B) services is understanding that special education and related services are no longer clinical in their focus, they are now directly related to the child's ability to obtain reasonable benefit from his/her education…that is, the focus is on an "educational disability."

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Special Education (continued from previous page)

In preparation for a referral conference, it is in the child's best interest if parents ensure that any and all information is submitted to the LEA that will assist in determining whether or not the child has a disability. This includes, but is not limited to, the results of hearing and vision screening; home or classroom behavior checklists; existing medical, social, or educational data; examples of the child's academic work; and screening inventories. Most of the time, only some of the people attending the referral conference have an adequate knowledge of your child. Considering that these documents can "make or break" your case for having your child evaluated for special education and related services, this supportive documentation's importance cannot be stressed enough.

Additionally, IDEA requires that a referral conference must occur within a specified amount of time after having received a request for referral. In our state, the school has seven days from the written request to schedule a conference at a time and place agreed upon by the parents and provide the parents with written notification of the referral and a referral conference notice. If the parents do not respond within seven days of the notification, then we allow another seven days and reschedule for what is now 21 days from the initial referral request. The referral conference transpires on that date regardless of attendees. The short of this story is that if you request a referral, expect it to be held within fourteen days from your request. If you need more time to compile your documentation or ensure attendance by someone you have invited to the conference (e.g., an advocate or medical professional), then let the school know; however, be aware that the conference must occur within 21 days of the request despite scheduling conflicts, postal difficulties, fax malfunctions, etc. on the parents' part. Again, I am assuming that other states have the same timelines, but check with your state department of education if you have questions or concerns regarding this.

At least three people must attend the referral conference. Usually it is a teacher involved with your child, a parent, and a school administrator. Upon completion of the conference, then written documentation regarding the conference decision is compiled and a copy is provided to the parents at that time. If the parents are not in attendance, then it is provided to the parents within seven days.

If the decision of the committee is that a comprehensive evaluation is warranted, then you must give written informed consent for the LEA to conduct it. Generally, you will be asked to complete a social history and provide a few other pieces of information, if necessary, at that time, too. The decision may also be that a specialized evaluation is necessary. It may be that your concerns only relate to your child's speech or language development. This is a specialized evaluation because it doesn't require all of the components of a comprehensive evaluation in order to determine if your child displays characteristics of an educational Speech or Language Impairment. Again, informed consent is required. Finally, the decision may be that no evaluation is necessary. This could occur when supportive documentation is not provided or complete, the child has a history of excessive absences or moves from school to school, or the child has sensory issues (e.g., needs glasses) that must be addressed and could explain why the child is experiencing academic difficulties. If the decision for a comprehensive evaluation also includes a temporary placement, then this placement is limited to 60 calendar days and evaluation must occur within the same time frame.

In conclusion, parents don't get the chance to tiptoe or wade into the demands of raising a child with special medical and educational needs. I know it felt like a sink or swim scenario for me. My daughter's medical issues were the hardest part for me to undertake, initially. I was consumed with just trying to keep her alive. When she was able to be enrolled in a facility that provided day habilitation, therapy services, and medical care during the day, I thought I might feel a bit of relief. Little did I know that my knowledge of special education actually made it more difficult to be a quiet advocate for her. I kept discovering things that the facility was doing that were in violation of state and federal rules and regulations regarding IDEA and health insurance. Jeez, now I was constantly in battle with a nationally known agency that served children and adults with special needs. My plate runneth over! She is in second grade nowadays and I realize everyday that things happen for a reason and that every experience, good and bad, prepares you for another one. In many cases, we are the only voice our child has. Kudos to those of you who get out of bed and put your "advocacy armor and shield" on everyday. Our visiting angels may only bless us with their presence for too short of a time, but each and everyone one of them enriches our lives in ways that we may only realize far in the future. Two of my favorite quotes will summarize my thoughts on all of this. Please enjoy:

"You gain strength, courage and confidence by every experience in which you really stop to look fear in the face." ~Eleanor Roosevelt

"If children have the ability to ignore all odds and percentages, then maybe we can all learn from them. When you think about it, what other choice is there but to hope? We have two options, medically and emotionally: give up, or fight like hell." ~Lance Armstrong

Cathie Acosta
Mom to Piper, age 7 - CblC, and Dillon, age 17.
29 Easterwood Point Rd.
Mayflower, AR 72106-9650

2010 OAA Shirts Available Now!!

Café Press has a large selection of items available with the OAA cover printed on them. They carry long and short sleeve shirts, tees, sweat shirts, bags, ornaments, license plate holders, toys, mugs, magnets… and calendars galore. Be sure to check out the pictures on the insides, too.

http://www.cafepress.com/organicacidemia

Nutrition Guidelines Project: OA Workgroup Focus on Propionic Acidemia (PROP[1])

Project Primary Investigators: Rani H. Singh PhD, RD & Frances Rohr MS, RD
OA Workgroup Chairs: Elaina Jurecki, MS, RD & Keiko Ueda MPH, RD
OA Workgroup Members: Nancy Baugh MS, RD, Laurie Bernstein MS, RD, FADA, Lisa Bingen MS, RD Christie Hussa RD, MBA, Ann Marie Roberts RD, CSP, CNSD, Heather Saavedra MS, RD, Steven Yannicelli PhD, RD

The development of nutrition guidelines for PROP is needed as there are multiple approaches for the nutrition treatment of patients living with PROP but no clear consensus on best practices to promote outcomes[2,3]. PROP due to the non-working propionyl Co-A carboxylase gene, is one of the rarer inborn errors of metabolism (IEMs), with an estimated incidence of 1:100,000[4]. With 4.1 million births per year in the US, there are approximately 41 infants with PROP born per year[5]. Despite the fact that PROP was first described over 50 years ago in 1961[6], there is still much that we do not know about the natural history of PROP. Medical nutrition therapy (MNT) is a key therapy for patients living with PROP but there are many reports of poor neurologic outcomes. There remains a high rate of morbidity and mortality from either cardiomyopathy and/or from episodes of metabolic instability due to intercurrent illnesses[6].

Due to the rarity of IEMs, there is a lack of published evidence-based literature standardizing the MNT of any of these disorders[8] including PROP[2,9]. The Southeast newborn screening and Genetics Collaborative (SERC[10]) funded through the Maternal and Child Health Bureau Health Resources and Services Administration (HRSA), Department of Health and Human Services o administered the Nutrition Guidelines Project in collaboration with Genetic Metabolic Dietitians.(GMDI) to address this national gap. The purpose of the project is to develop nutrition guidelines for the management of genetic metabolic disorders where there is limited published scientific evidence. As a pilot five workgroups were formed to represent the following groupings of IEMs: aminoacidopathies, fatty acid oxidation disorders, organic acidemias (OA), phenylketonuria and urea cycle disorders. PROP was chosen as the first disorder to focus the efforts of the OA workgroup based on a GMDI metabolic dietitians' needs assessment survey; other workgroups are focusing on maple syrup urine disease (MSUD), Phenylketonuria (PKU), and validating the previously developed nutrition guidelines for medium chain acyl Co-A dehydrogenase deficiency (MCADD) and very long chain acyl Co-A dehydrogenase deficiency (VLCADD). Workgroup chairs were chosen by project primary investigators based on their experience in the field. A core group, including the primary investigators, study coordinators and workgroup chairs, oversee the progress of the workgroups. The OA workgroup includes nine metabolic dietitians with a combined 102 years (average 11 years per dietitian) of clinical IEM patient experience. Dietitians were recruited from the professional group, Genetics Metabolic Dietitians International (GMDI)[11]. A nutrition guidelines project website was developed to facilitate workgroup member communications and the systematic sharing and editing of documents.

The OA workgroup summarized a list of 74 questions to address the unanswered or ambiguous MNT questions for PROP. The ultimate goal is to develop a standard of care for the MNT of PROP. The four main areas of MNT to establish consensus include:

1. Establishing the dietary amino acid and nutritionl goals when starting diet therapy,
2. Illness nutrition management guidelines,
3. Impact of biotin and L-carnitine use on MNT,
4. Optimal nutrition therapy for patients living with PROP and cardiomyopathy, diabetes, pancreatitis or receiving a liver transplant.

A systematic and transparent process was developed to review published medical literature and to identify and address IEM specific MNT concerns utilizing a combination of evidence based analysis[12] and consensus methods. Evidence-based analysis methods provide workgroups with unbiased and critical assessment of the quality and relevance of published literature for MNT in PROP. Published literature in MNT of PROP is limited. For this reason, the workgroup will also solicit feedback on common clinical practices from expert metabolic physicians and dietitians chosen from the seven US HRSA regions[13]. When gathering clinical practices the workgroup will use established consensus methods such as delphi surveys and nominal group meetings[14]. Delphi surveys will be mailed to participants and include PROP MNT clinical practice statements that invite comments. Moderated face-to-face nominal group meetings will be held to clarify ambiguous MNT practices. The consensus techniques will invite comments from expert metabolic dietitians, physicians, other healthcare providers and patient and family community stakeholders. The goal is to incorporate feedback from a large group of health care providers who are taking care of PROP patients. The PROP and other IEM nutrition guidelines will be field-tested and once finalized will be published on the SERC and GMDI websites and subject to future revisions.

Developing a consensus on nutrition therapy for PROP and nutrition guidelines for other IEM's is timely given the expansion of state public health newborn screening programs. These programs identify patients who require diagnosis and treatment as early in life as possible. Development of an evidence-based and consensus-validated PROP medical nutrition guideline will help to: standardize PROP MNT, improve consistency in PROP nutritional therapy and management, allow for future PROP guideline revisions, and support the main goals of improving the quality of medical care and patient outcomes. A PROP nutrition guideline will focus future directions in MNT, medical and nutrition professional and patient continuing education, and clinical nutrition research for PROP.

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Angels for Alyssa/MMA Research Fund

The Angels for Alyssa/MMA Research Fund is focused on financial support of scientific research towards a cure for MMA being performed by Dr. Charles Venditti at the National Institute of Health (NIH). MMA donations can be sent to:
Angels for Alyssa/MMA Research Fund
c/o Marty Moran
PO Box 262
Shermans Dale, PA 17090

OAA Research Fund

The Organic Acidemia Association is pleased to announce a new research fund dedicated to research that should eventually help all Organic Acidemias. Currently we have a contract supporting Dr. Charles Venditti at the NIH but we may choose to support other researchers if we feel they qualify. OAA donations can be sent to:
Organic Acidemia Association
c/o Kathy Stagni
13210 – 35th Avenue North
Plymouth, MN 55441

An Update on Charlene Maloney, GA1

For those of you who do not know us, my name is Debbie and I am the proud Mom to Charlene, 17, GA1. Char was diagnosed at 8.5 months old after a metabolic stroke. This left Charlene absolutely listless. She lost all of her motor skills, including being able to drink a bottle. Char went through many years of struggles and surgeries.

In August of 2005, Charlene had a tracheotomy at the AI DuPont Hospital in DE. She was put on a ventilator at this time. After this surgery, it took Char a long time to recover. The following summer we took her to see her favorite country singer, Trace Adkins in Nashville TN. I noticed Char's breathing was better and started making plans to move here! We moved in August of 2006 and have been here since.
Charlene is still the happy go lucky girl she has always been. She has not needed any more major surgeries. Char has been off of the vent for about 2 ½ years now. She is a junior in high school and loves it! She has wonderful nurses who accompany her to and from school.

Charlene's major issues lately have been bed sores. She had a really bad one at the bottom of her spine, which is now closed up. On our last appointments with her pediatrician, neurologist, and plastic surgeon they told us there is no need to keep coming back unless we have an issue! Her pulmonologist said we can see him annually instead of every six months! She is doing excellent in all areas! Of course Char still has no real motor skills to speak of but she can laugh, smile and enjoy life like no one I know!

I am curious as to any children older than her in the same condition. I just do not know what to expect next. Char has had a baclofen pump for almost ten years now. This has greatly helped with the spasms! She has a feeding button, trache, has almost no motor skills, and cannot talk.

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Organic Acidemia Association
13210 – 35th Avenue North
Plymouth, MN 55441

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 501(c)(3) non-profit corporation. Donations to the OAA are tax deductible. OAA publishes a newsletter 3 times a year, hosts a Google Group for information exchange and maintains a website. Services are funded by corporation & individual membership 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.

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 OAA Group. Visit the OAANews.org web site to sign up. This is a private list not open to the general public (but you never know who may be "lurking").

* 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.

Facebook
OAA is on Facebook -- donations can be send through our "Cause" Page, connection with other parents can be found through our "OAA Group" and "Fan" Page.

Wanted
* Articles are ALWAYS needed for the newsletter.
* We will be needing volunteers to help at the conference in July.
Email if you're interested in helping!