



Organic Acidemia Association

NEWSLETTER

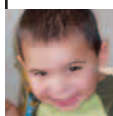
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Happy Holidays!

It's hard to believe that yet another year has passed and soon it will be 2006. A lot has happened in the past few months since the last newsletter. In this edition of the OAA Newsletter, you will find it jam-packed with pregnancy, press, partnerships, people, and of course politics. You won't want to miss ...

The very informative article by Drs. Mardach and Cederbaum discussing Cardiomyopathy and its impact on those with Propionic Acidemia. The doctors explain how they discovered a link between this deadly heart condition and patients with PA. As a PA mom I found this particularly interesting since Melissa's heart has been routinely monitored. To my surprise, recently she was found to have a mild case of Cardiomyopathy and is now on medication to lower her blood pressure in hopes of preventing further damage to her heart.

In the news, we have been asked to introduce OAA families to the Human Genetic Cell Repository where your contribution of a blood sample can help researchers today and tomorrow as they work on improved treatments and potentially a cure for inborn errors of metabolism.

On the research front, the OAA has launched a new research fund for Propionic Acidemia. The OAA/PA research fund will work just like the OAA/MMA research fund with tax-deductible contributions collected by the OAA and then distributed to approved researchers by fund trustees.

In a related note, I have recently resigned my position as secretary and member of the board of directors of the Propionic Acidemia Association (PAF). My decision to disengage with the PAF was the result of a continuing disagreement with the board regarding the focus and mission of that organization.

The next national family conference is still on schedule for next summer in Dallas Texas. We are still looking for volunteers who can help with pre-event coordination as well as assisting during the conference. If you are interested in helping, please contact me directly through the OAA phone/email.

And as if all of this is not enough, look for some big changes in the OAA Internet web site. We are near completion of a major site upgrade and plan to move to the new site by the end of this year. Check out the special review of the new site inside the newsletter.

Finally, it is that time of the year to ask for your continued financial support of the OAA. We recently sent out reminder post-cards to members asking that they renew their membership for 2006. The OAA continues to offer more and more information and support services, and the cost to deliver these are increasing. Postage rates go up in January. Our new web site will no longer be provided at no charge increasing our operating costs. To keep pace with these rising costs, we have increased the requested (voluntary) annual membership fee from \$25 to \$30. Your continued support ensures our continued ability to support you!



2006 FOD/OAA National Metabolic Family Conference Dallas, Texas June 23-24, 2006 ~ Hosted by ~ The Institute of Metabolic Disease Baylor Health Care System Dallas, Texas

Location: Adam's Mark Hotel 400 North Olive Street, Dallas, TX 75201

Call for Hotel Reservations: (214) 922-8000 (each Family makes their own reservation) <http://www.adamsmark.com/dallas/index.asp> **Special room rate: \$ 95.00 (single/double)

You must state that you are attending the FOD/OAA National Metabolic Conference hosted by The Institute of Metabolic Disease

Transportation to/from the airport (DFW and Love Field): (Each Family must make their own reservation on the Yellow Checker Shuttle website (www.yellowcheckershuttle.com/jeff) in order to get the discounted price of \$13.00 one way - or book your own company choice)

- June 22** Welcome Reception/Cocktails, 6:30 - 11:00 pm at Hotel
- June 23** Metabolic Conferences (Each Group in own room), 8:00 am - 5:00 pm
- June 23** Dinner on own, Good time for Family Networking 6:00pm on
- June 24** Combined FOD/OAA Session, 8:00am – 12:00 noon
- June 24** Tour of Institute of Metabolic Disease 2 – 4 pm (van provided)

Coming Soon Redesign of OAA's website

www.oaanews.org

(see page 13 for web picture)

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 listserv for information exchange and maintains a website. These services are funded by donations from corporation and individual members. Annual membership donation of \$30 (U.S) and \$40 (international) plus \$5 for the family roster is requested, but not required. Our 501(c)(3) non-profit status qualifies OAA for United Way donations through their write-in option. If there is a write-in option, just write "Organic Acidemia Association" in the blank line on your pledge card. Donations can also be made at OAA's website through the "PayPal" and the "Network for Good" option.

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

Make a Contribution Today and Be Part of the Solution Tomorrow

The Human Genetic Cell Repository (HGCR) at the Coriell Institute for Medical Research is currently seeking volunteers to donate a blood sample for research. The HGCR, which has been sponsored by the National Institute of General Medical Sciences at the NIH since 1972, serves the research community by collecting, storing, and distributing cells from individuals with genetic diseases. Qualified researchers can obtain these cells to study how cells function, to identify new mutations, and to develop ways to diagnose, treat, and possibly prevent genetic disease.

Due to the expansion of newborn screening in the United States, the HGCR has undertaken a project to obtain samples from all 29 disorders on the newborn screening panel. The increase in the number of disorders tested for via

newborn screening will generate new research initiatives to better understand these disorders. With your help, the HGCR can help provide the appropriate materials needed to accomplish this research.

Any individual with an inborn error of metabolism, such as an organic academia or fatty acid oxidation disorder, is eligible to participate. A blood sample and clinical information about the donor is all that is required. All samples and information will be anonymized and no identifiable information will be released to researchers. For more information or to sign up to donate, please contact Tina Sellers, MS, Genetic Counselor for the HGCR at (856) 966-5062 or tsellers@coriell.org.

Special Education Services IDEIA 2004 OVERVIEW

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Students who receive special education services are starting the 2005-2006 school year under a new federal law. The Individuals with Disabilities Education 'Improvement' Act [IDEIA], signed into law in December, 2004, took effect on July 1, 2005. The federal regulations implementing the new law have not yet been finalized. The reason Congress gives for changing the federal law was to align it with the No Child Left Behind [NCLB] Act. State special education rules remain in force where and when they provide more benefits or protection to the students than IDEIA will.

The changes made by IDEIA touch on all areas of special education law. IDEIA adopts the NCLB's "highly qualified" requirements and applies them to special education teachers. IDEIA ensures that Individualized Family Service Plans [IFSP] and Individualized Education Programs [IEP] use methods and techniques that have a track record of success. IDEIA requires that the IFSP/IEP services focus on scientifically based approaches that have been tested and reviewed by educational professionals.

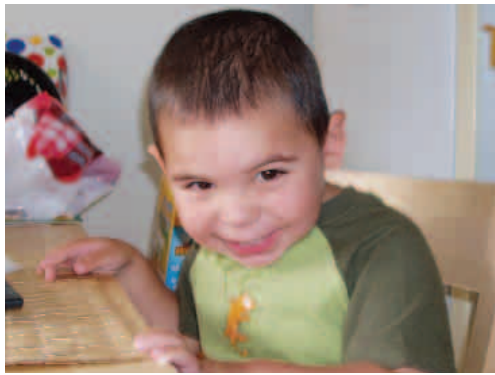
The scope of parent participation is strengthened in some areas and weakened in others. There will be new procedures used to

regulate who must attend Team meetings and how IEPs are prepared. IDEIA also permits IEPs to be amended.

IDEIA changes the age at which schools must begin to consider student transition from the classroom to the community. The new age is 16. IDEA '97 started the transition process at age 14. IEP transition plans must place greater focus on skills that are vital to a student's success in the community.

IDEIA changes the due process system used to protect a student's right to receive educational services. When parents request a due process hearing a new step is added. From now on, parents will have to meet with school officials in a "resolution session" before going to a due process hearing. IDEIA gives specific qualifications all hearing officers must meet.

Student disciplinary procedures will change under IDEIA. Schools must consider a student's challenging behaviors when writing or revising an IEP. If a student is being considered for disciplinary action, new procedures are used to determine whether the student's conduct is caused by the student's disability. Parents will need more proof to connect the student's conduct to her or his disability than was required by IDEA '97.



Gabriel Lopez, Propionic Acidemia, Age 3

Hi everybody. We are delighted for the opportunity to share our story with the OAA family, hoping that it may help other families to go through a diagnosis of a metabolic disorder.

My son Gabriel was born in London 3 years ago. Being our first, long-awaited child, we were ecstatic when he arrived. He was gorgeous! The main thing I noticed when Gabriel came out was that he didn't cry. That was odd to me but nobody seemed particularly worried about it since his APGAR scores were excellent. To this date, we still don't know if he was already in trouble the minute he was born. The only "real" problem that he had at birth was that he was very big, and his blood sugar was a bit low. I immediately tried to breastfeed him, but he nurses thought he hadn't had enough and encouraged him to take his very first milk bottle.

I was very eager to breastfeed Gabriel but the first night of breastfeeding was exhausting for both of us, as Gabriel would take at least one hour and a half each time, and he wanted to eat every two hours. In addition, he didn't seem to cry like the other babies in the ward (there were 10 beds per ward and all of the women would breastfeed). Being a first-time mom I thought this was quite tough and exhausting but decided that I needed to put more effort into it.

The second night was even worse. I said to the nurses that he was grunting a lot all the time and the nursing was not really like the other babies. They ignored me completely (several of them through the night), and suggested that it was normal or that I didn't have enough milk for the baby. In the morning, Gabriel finally fell asleep in my arms after a long night of trying and trying to breastfeed. I put him in his cot to rest. When the morning nurse came to take my blood pressure, I said to her that the baby didn't seem happy breastfeeding. She then noticed his grunting and that his chest was moving up and down very fast (something the other nurses didn't care about). I will always remember the name of that nurse, Marcy, a nurse that came from Gambia (West Africa) for training in the UK. She immediately called the pediatrician from the NICU unit, who came right away and decided to take Gabriel away for further observation. That was the beginning of our journey with an organic acidemia.

We visited Gabriel a few hours later at the emergency unit and he would still look gorgeous (all of the babies down there were premature, really, really tiny!). No-one would have said that he had been very acidotic and that his ammonia had been quite high. The NICU doctors did a wonderful job stabilizing Gabriel very quickly and bringing his ammonia down. They said that a few more hours would have been fatal. The pediatrician in charge then asked me if I had taken cocaine or other drugs that could explain Gabriel entering into such a deep crisis. After the results of most of the tests came back (spinal tap, EEG, blood work, urine, heart, lungs...), they suspected Gabriel might have something metabolic and contacted the metabolic unit at Great Ormond Street Hospital for Children, a hospital that specializes in rare diseases and was (luckily!!!) a few blocks from where we were.

They ordered to test for urine organic acids and to have Gabriel transferred to their unit. That afternoon, the doctor broke the news of a possible metabolic disorder to us at the maternity ward. We were really shocked! We went home with a deep sadness and returned the next day after to discover that Gabriel had already been transferred to the metabolic ward. We met with his doctor, Prof. Leonard, a very knowledgeable and kind man, who was so admired and respected by the fellows, students and nurses. A few months later we understood why. Prof. Leonard said that we needed the result from the organic acids but that they suspected, on the basis of the pattern of blood amino acids, a diagnosis of propionic acidemia. He also suggested us to go to the web and find out information about it, read it and come back the day after with questions. We were still in denial and shock and to read all that information in the middle of the night with the empty crib in our room didn't help at all.

The days passed and Gabriel started recovering slowly, they removed the IV, put an NG-tube and started giving him a mixture of sugar (Maxijul, the British equivalent of Polycose) and baby milk. They would add a bit more milk everyday to see how his tolerance was. Later on, that was substituted by breast milk. And a bit later on, the NG-tube was removed and I started breastfeeding. Now, he was a much, much happier baby than at birth. What a change!

We were sent home after 15 days in hospital and with a big bunch of papers and emergency protocols and information in case Gabriel happened to get sick. Two months passed and Gabriel seemed to be growing very well. At two and a half months, his first vaccination took place: a combination of 5 vaccines given in two shots, all the same day. A few days later Gabriel got into

trouble: he started vomiting very violently in the middle of the night. He looked so pale that I thought he was dying! We rushed to the emergency protocols and started giving him sugar for a few hours. He seemed better and 24 hours later he started recovering. But then, I noticed that he started sleeping a lot everyday, especially in the mornings. At our monthly clinic that Friday nothing unusual was noticed. Prof. Leonard was happy to see what we had done and no special adjustments were made. However, I noticed Gabriel was not quite the same. Another week passed and I realized he had not been smiling anymore since his vomiting episode. I called the metabolic ward to see if I could do anything. After consultation, the nurse that had helped Gabriel during his first crisis said to keep an eye and call back if he vomited again. That never happened and I didn't call back (I still wonder why I didn't do it).

Days later Gabriel was also getting constipated. He would not wake up to nurse, he wouldn't nurse in the morning and then at night he would want to breastfeed to much. He was again grunting. When I mentioned all his sleepiness to the nurses and pediatricians of our regular visits, they would say it was quite normal for young babies to sleep a lot, not to nurse too much, and not to be very happy when they were constipated. When my husband came home from work in the evenings, Gabriel would start being very awake again. So he would find him actually quite well.

Two more weeks passed and another time for vaccination arrived. After that, Gabriel got even worse, he would still fix his eyes and looked like a nice baby but somehow he wouldn't put any weight on. Nobody thought he was sick (he was a "good baby"), but I was still worried since he didn't seem to be too active, still very sleepy. And he still didn't smile. When Prof. Leonard saw him at the metabolic clinic (two months after the first immunization), he immediately noticed that something was not quite right. Gabriel was excessively floppy and he was not happy at all. He was admitted to hospital two days later. After all the proper tests were done, we found out Gabriel was not very stable metabolically. But he wasn't terribly bad! He was stabilized with IV glucose and started him back to milk in a few days. They concluded he was not having enough milk with the breastfeeding and prescribed 1 L of milk/day. He put weight very rapidly (he grew in three weeks the equivalent to three months!) However, something else had to explain the floppiness that he still had and the lack of smile.

An MRI was ordered and the result was inconclusive. Then an EEG was ordered and the

continued on page 4, Gabriel Lopez

result shocked us, as Gabriel was showing an amazing amount of abnormal brain activity (hypersarrhythmia). A neurologist visited Gabriel and he prescribed anticonvulsants right away although Gabriel had never had a visible seizure before. The first drug, carbamazepine, had effect four days later, although it wasn't the effect we expected: Gabriel started having fits. He would roll his eyes a bit and lift his two arms slowly. This would last for some seconds and stop. First, he would do it for two times a day, then four, then eight and every day he was having more and more episodes. It was so horrible to watch! His smile was still not back. The neurologist then added phenytoin. This drug changed the seizure pattern, making them a bit more frequent, but there would not be any improvement.

At that time, after three weeks in hospital and with Gabriel totally stable metabolically, we were sent home. The doctors wanted to think about what to do, but they didn't think Gabriel had to stay in hospital. He was taking his liter of milk nicely from the bottle and we went home two days before Christmas having cancelled all our holiday arrangements. Gabriel looked much healthier but was still having lots of fits. Now, he would cry after each episode, as he was beginning to be conscious that something bad was happening to him. After the holidays, we wanted to stop all the medicines, as we didn't see any positive effect. Prof. Leonard agreed and suggested another neurologist from the hospital: Dr. Helen Cross, a specialist in child epilepsy. We got an appointment three days later. After explaining Gabriel's behavior and history to her, she immediately diagnosed him with infantile spasms, a rare form of child epilepsy with high chances to cause developmental delays and more epilepsy later in life. She put him on vigabatrin and started to wean him from the other anticonvulsants slowly. If nothing happened after three days, she would increase the dose. If still nothing happened, she would change the treatment to steroids (something no-one was happy about because of all the metabolic implications).

I cannot tell you how miraculous that drug was. After 24 h on vigabatrin, Gabriel stopped completely having fits. After 48 h, he started smiling again. After three months, it was such an unbelievable day! I took so many pictures! Gabriel continued improving by the day. He would start developing and recovering quite fast. We didn't have to use the full dose of the drug since he responded so well. Things continued improving all the subsequent months, although Gabriel was developmentally quite delayed and all his milestones were not achieved on time.

Coincidentally to the effect of the vigabatrin, Gabriel started drinking less and less formula every day and a nasogastric tube was put in place to ensure he would get all his formula requirements.

We had introduced solids at 6 months of age when we left hospital and, although he was initially very happy eating them, his consumption of solids was also declining by the day. Every time we had to feed him, it started to become a huge struggle. That winter, Gabriel was admitted to hospital approximately every two weeks. First with RSV infection, then asthma-like symptoms, then another cold... I was losing so many days of work to be with him at hospital!! Luckily, we went always home after two nights. His ammonia levels recovered quite quickly every time, and he would stop vomiting after 24 h of protein-free formula. Also at 1 year of age his dietitian changed him to a new formula based on Maxijul, vitamins and Pediasure. The change from baby milk to Pediasure dramatically reduced the constipation problems he had had in the past (who knows why).

At 12 months of age, Gabriel was weaned off the vigabatrin. He never showed signs of epilepsy again and he was still progressing, although quite slowly. At the same time, by 15 months, a developmental pediatrician evaluated him and, although Gabriel was at the low end of the curve, the doctor wanted to give him a chance to develop by himself now that he was doing so well metabolically. Unfortunately, Gabriel suddenly stopped developing and, by 18 months, he had not made any progress from the previous visit. He didn't look unhappy, sick or epileptic; he just wouldn't do anything new. Therapy was recommended but we had planned to move to New York two months later so therapy had to wait.

At around that time, his NG-tube was replaced by a G-tube. That month, Gabriel stopped eating all solids because he would have difficulty swallowing anything. We got scared and stopped also offering any food. The problems related the G-tube that we encountered during the first month (lots of reflux and vomiting) resolved slowly. We found out that Gabriel tolerated much better his feedings if he seated at an angle. His stroller was perfect for this. Feeding therapy was on hold because of our moving to the United States.

When we arrived in New York, and entirely out of the blue, Gabriel started developing again. He would even start some word approximations. At 18 months of age he was not walking yet and wouldn't do it for another 6 months. However, he started to have therapies with the Early Intervention Program and that made a huge difference. Unfortunately, three months later his speech regressed and never developed beyond making a few sounds. Although Gabriel looked quite happy overall he would be very silent for most of the day. In terms of hospital visits, he was admitted at Mount Sinai for minor infections and had to stay in hospital for several days after a change in diet made him very sick. After that, more than a year ago, Gabriel has remained out of

the hospital. We have learned to manage him at home every time he vomits or looks inactive, sleepy or just strange, following the initial emergency protocol we were given in London when Gabriel was first diagnosed. That has worked every time during all these three years (20% Polycose, administered at a rate of 60 mL per hour).

Gabriel travels often to see his grandparents in Mexico and Spain (he's been doing it since he was 2 months-old). He gets a bit sick because of the tiredness, but we just give him some of the emergency formula while and after traveling and that helps tremendously. He is a curious and active boy who loves to go to the playground and the Children's Museum in Manhattan. His major challenges are speech (he has made very limited progress despite all the therapy he's received) and hypotonia, but he is now doing very well and keeps progressing at a steady pace. He goes to clinic once every six months to Children's Hospital of Philadelphia, followed by Dr. Paige Kaplan, and despite getting ill with common infections he has never been acidotic again. He is fed 100% via the G-tube with a formula that contains Polycose, vitamins and Pediasure. He takes 5 cc of carnitine twice a day.

We would love to see Gabriel eating by mouth one of these days and we are trying very hard with different strategies (oral/motor and sensory integration techniques). He has received intensive therapies at home for a whole year with the Early Intervention Program but now he just started in a special education preschool program with 2 x OT, 3 x ST and 2 x PT. We are all very excited about school, as we feel he desperately wants to interact with other children. He is making progress by the day and although still functions with delay he is a sweet and lovable boy. When we started him on oral/motor therapy, he started making many more sounds and some word approximations in all three languages he hears (Catalan, Spanish and English). I don't think he's ever been so happy! A recent 24 h EEG showed no traces of any abnormal activity in his brain at all. Recently, Gabriel is finding a lot of joy with his little brother, Alex, born a year ago and tested for propionic acidemia with Newborn Screening (through the Pediatrx test). Alex is unaffected. We hope this year continues to be as amazing for Gabriel as has been so far. He is so eager to play and learn and enjoy life. We totally embrace his happiness and hope it will last for many years to come.

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Cardiomyopathy in Propionic Acidemia

Rebecca Mardach, M.D. and Stephen Cederbaum, M.D.*

Approximately fifteen years ago, a newborn infant was transferred to the neonatal intensive care unit at UCLA following a four day history of continuous deterioration after a normal pregnancy and delivery. A diagnosis of propionic acidemia was made fairly quickly and what is now recognized as appropriate therapy was instituted.

Like so many patients with propionic acidemia, the patient was comatose, had very poor muscle tone and had difficulty maintaining blood pressure. Because we had become so accustomed to assuming that the low blood pressure was due to the neurologic effects of propionic acidemia, it did not enter our mind that there could be any other cause for this blood pressure problem. Fortunately, the resident in the neonatal intensive care unit was unburdened by any such assumptions and evaluated the low blood pressure as he would blood pressure in any sick newborn. Part of this evaluation involved obtaining an echocardiogram of the heart, a procedure which looks for structural abnormalities but also determines the output of the heart and its ability to contract effectively. The study revealed that the low blood pressure was almost certainly due to the very poor ability of this patient's heart to contract. The resident assumed that this was an independent condition causing severe cardiomyopathy.

It immediately occurred to us, however, that there was probably no reason why a compound such as propionic acid, which can intoxicate the brain and the bone marrow, could not intoxicate the heart and cause it to function less efficiently. We reasoned, as it turned out correctly, that there was no need for an independent evaluation for heart disease, but rather the condition would clear up as soon as the acute propionic acid intoxication cleared up. This turned out to be the case, and a follow-up echocardiogram revealed an entirely normal cardiac function.

Because this idea was then a departure from commonly held beliefs, we shared our thoughts with several colleagues, all of whom agreed that this was a perfectly reasonable idea. Some, in fact, shared similar experiences and thoughts, although nothing had appeared in the medical literature at that time. From then on, we remained alert to the occurrence of inadequate cardiac output in propionic acidemia. One group's experience was

formalized and reported in 1993 when Massoud and Leonard reported 7 patients with propionic acidemia who had a cardiomyopathy, either acutely during an episode of deterioration, or of more gradual onset, without an obvious metabolic trigger.

Shortly after this, a young 8 year old girl with propionic acidemia for whom we were caring, and who had been under scrutiny for cardiac problems, was admitted to the hospital with a new onset hypertrophic cardiomyopathy (poor cardiac function in a heart whose muscles had grown and expanded to compensate for inadequate function). The size of the heart indicated that this had been going on for at least several weeks. Unfortunately the process was very severe and far advanced and the patient died shortly after she came into the hospital.

“It occurred to us that there was no reason why a compound such as propionic acid, which can intoxicate the brain and bone marrow could not intoxicate the heart and cause it to function less efficiently.”

We were so shocked and dismayed that this patient with a relatively mild form of propionic acidemia had developed so severe a fatal disorder so quickly and right under our noses, that we determined to try to learn as much as we could as to why this occurred. A complete autopsy was done and the heart was studied biochemically. The results suggested a number of things. Despite normal plasma carnitine and carnitine supplementation, the levels of the carnitine in heart were reduced. Furthermore, they suggested that one of the steps involved in the generation of the energy needed to keep the heart running effectively was impaired.

The results of our studies were the product of a unique opportunity and pointed to a problem with energy metabolism. The exact cause of the cardiomyopathy could not be pinned down from this one study alone, but

they do point the way for studies that may need to be done in the experimental mouse model of propionic acidemia. Before any thought of treatment or prevention can be undertaken, we need to know if these results are likely to be general and need to know what might affect the process in an animal model.

The study did, however, reaffirm the fact that patients with propionic acidemia appear to be predisposed to cardiac difficulty and although our understanding of the appropriate interventions are more limited, the importance of being aware of this and following patients closely for the possibility of cardiac deterioration is clearly necessary.

How then do we feel that patients with propionic acidemia should think about cardiac disease? Firstly, it is important to bear in mind that many; if not most, patients with propionic acidemia will not develop cardiac disease or life threatening cardiac disease. Secondly, many of the problems that might develop are treatable and manageable and shouldn't be viewed as a terrifying prospect. Our own practice is to have every patient with propionic acidemia followed regularly by a cardiologist. We recommend that they be seen on a yearly basis and have an echocardiogram at least every other year, if not yearly. We recommend a yearly electrocardiogram or failing that at least of a recording to make sure that the cardiac rhythm is regular and not beating abnormally. In the absence of specific guidelines, any abnormality should be treated as would the same abnormality in a patient without propionic acidemia. We certainly treat patients with carnitine and try to maintain free carnitine levels in the middle of the normal range at least.

We certainly believe that “an ounce of prevention is worth a pound of cure” and that we can prevent some otherwise serious problems in this way.

* Rebecca Mardach is Director of The Regional Metabolic Center at Southern California Kaiser Permanente and is an Assistant Clinical Professor in the Genetics Division of the Department of Pediatrics at UCLA. Stephen Cederbaum directs the metabolic program at UCLA and is Professor of Psychiatry, Pediatrics and Human Genetics. He is a new member of our medical advisory board.

Hannah Vogt, Glutaric Acidemia, Type 1, Age 4

Hi everyone! This is part of our family's story. I have prayed for a little girl since I was 12 yrs. At 30, I finally met and married Brad. We were so excited to be pregnant with our first child(ren)? They, however never developed in my womb. The doctors called this a blighted ovum. My body wouldn't miscarry my precious children, so the doctor wanted to do a D&C. I made her wait for 6 weeks or more until I was sure that the baby(s) were really not developing. It was Christmas time, and I was a workaholic, (before my injuries and illnesses, that came after Hannah was born.) The doctors told us we had to wait another year to get pregnant. So we waited. The advice seams bogus now.

Then the Lord blessed us again with our "precious angel", Hannah. I did everything I could to make the pregnancy perfect. I didn't drink caffeine. I made sure each meal I had all the food groups, etc... I was scared the whole pregnancy. I bled a little, due to placenta previa that corrected itself. Hannah tested positive for Down syndrome, by a triple screen test. Then, I saw a specialist and everything seemed alright.

I remember that I wanted to play a more active role in serventhood towards family's and children with disabilities. After Hannah was born, I tried to find ways to help out with Down syndrome kids and their family's, but couldn't seem to find any ways to do so. Isn't that sad? I wish that others would have easier access to knowing how to help with all of our situations. Hopefully getting Hannah's story out will help spark compassion, resources, and community, and of course complete pre-born screening in all 50 states. :) As well as, love, and hope, for those living with disabilities. It is funny that I wanted to help before we knew Hannah had GA1.

Finding out about her illness was a very long road, which, I will try to share briefly. At 1 day old Hannah could hold her head by her self. She was very strong the doctors said but she was very fussy from the pain she had. She was very beat up from a very traumatic birth that was uncalled for and disabling for me. I am permanently disabled from the injuries I sustained from her traumatic delivery. Her first week, Hannah turned blue and aspirated on day 2 and lost around 6 lbs. (?) Then Hannah became extremely jaundice. I

attributed it to birth injuries. After all, they ripped her out with forceps after other many aggressive things. We should have been offered a c-section! After longer than a week in the Hospital we came home. I cried and cried something felt wrong. I knew I wasn't post-partum, so I attributed it to my incredible pain and injury's. At 1 month or so I noticed problems with Hannah's head denting in. Then she lost some of her ability's. I wondered did I imagine she could do these advanced things. Then, her head, where her soft spot was, started rising up on one side, she was extremely fussy (beyond colic!) She would empty her entire stomach 5 to 12 x a day.(This happened for three years!!!) She couldn't sleep unless she was in our arms upright and we didn't move so much as twitch or we would awaken her to start a 3 hour or more ordeal again trying to calm her down enough to sleep and stop vomiting. At 2 months finally her head was so asymmetrical that our friend and doctor and huge lover of Hannah grew concerned so we had a CAT scan over a month later. The hospital read the CAT scan as "normal" with extra benign axial fluid of infancy. Which 2 or so weeks ago I found out that it clearly showed GA1 findings. Hannah was always screaming, non-sleeping, hungry baby. I felt she needed more than my breast milk, but when I would try formulas to supplement, she would really be vomiting. We tried many, many brands of formula, with no success. I thought the screaming threw the night was night terrors remembering her traumatic birth. I still wonder about that? At 4months (I think) she had a reaction to immunizations. After that well baby check I was complaining to our doctor that something seemed to be wrong as I thought again she had lost skills, and seemed like she was now not going to be able to sit up. (This was due to dystonia.) I was told that I was being a "perfectionist" and basically relax. I tried to explain without the proper terminology that she seemed like she would never be able to sit up. She also didn't have any protective reflexes. The doctor, Laura, said baby's usually sit up at 6months or so, and not to worry. So we waited till 6 months, yet Hannah still had no foreseen strength and ability to sit in my mind. 8months the same scenario was expressed, with don't worry. 10 months the same...so I took her to see my childhood



pediatrician. He said "it's good to be skinny," and she's "fine" "her constipation will go away," Not getting any answers became too much for me, so I heard about child find evaluations, and set an appointment. After many no shows they evaluated her and told me she had Hypertonia with possible cerebral palsy. Than finally, our doctor listened and set an appt. for around 2 months later to see a child development doctor. (Why she didn't send us to a neurologist I will never understand.) This doctor diagnosed her with spastic C.P. While I was waiting for an appointment, I was desperate for answers. I went to many doctors, tried going to The Children's Hospital ER and tell them she fell, so they would do a MRI, but I was unable to successfully lie. Not that they do an MRI on the weekend anyway. WHEW!!! Now I know with her condition being mistaken for shaken baby syndrome that could have been a fatal catastrophe for Hannah. If I wouldn't of been there, with her, to listen to my God given instinctive responses to feed and hydrate her all day long, if I would of been suspected for child abuse, like so many of you were. (My heart goes out to you!) I believe foster care wouldn't have had those needed instincts, because Hannah has always seemed to deteriorate quickly. Finally we were given spastic cp diagnosis, and then sent to an idiot neurologist, who said low tone cp. We went to 4 neurologists, 3 geneticist's etc. I always questioned everyone, because they said Hannah was dismorphic, but isn't, and they would test her for disorders she had no symptoms of. They did swallow studies, EEG's, EKG'S, sleep studies - this is where she meets one of the doctors who discovered GA1, yet still no GA1 diagnosis. She had 2 more MRI's over the course of two more years. I just found out the MRI's were "text book" of GA1 findings, yet no diagnosis. I kept seeking answers for Hannah because I knew she

shouldn't be non-ambulatory, aspirating, fussy, tiny, big head, slow hair growth, puking the entire time child... But most importantly, I wondered if she had bleeding on the brain due to asymmetry. I instinctively knew her immune system was very weak. Therefore I was very protective of her. Being protective and advocating, and seeking answers made people (Therapist mostly) think I was crazy. Also the new friends I made in Denver fled when they realized Hannah was disabled. I was left with no one to believe me, or listen and support me. Not my husband, family, and friends, or professionals. This was crazy, especially since she was so sick and non-ambulatory. Finally, when Hannah was around 2.5, we set an appt. to see this, "incredible" doctor. After 2 attempts, and around 9 months, we were finally able to see this awesome neurologist who finally diagnosed Hannah right away. Her boss, the idiot neurologist, would always come in instead of letting her, Dr. Parsons, see Hannah. I believe he was covering his tracks. He after many questions to him about Hannah's possible seizures and weird MRI's gave her another wrong diagnosis of Perisylvian Fissure Syndrome. I said to him right there that the assumption wasn't correct because Hannah was already advanced in speaking, even if he didn't choose to listen past the hard articulation. In Sept. of this year, we saw Dr. Morton and he believes the 4 or so people with Perisylvian Fissure Syndrome are actually GA1 cases. Hannah was three when she was finally diagnosed. I'm extremely glad she is alive. :) !! We probably had her on around 36g of protein a day before we knew. She had extreme feeding issues, and still does, so back then we feed her soft mechanical foods to avoid choking, vomiting. The soft mechanical foods we used were high levels of protein like yogurt etc... It is actually a miracle that Hannah is alive and doing as well as she

is. Even though her disorder wasn't properly diagnosed, for almost 3 yrs., even when all the evidence was obvious! It is also a shame we were not told that for 25\$ we could have had a more expansive newborn screening for 27 additional Disorders here in Colo. I will be calling Pediatrix to order a kit for newborn screening that will test for about 50 disorders. This kit will be for my son Kyle. I believe, he has autoimmune dysfunctions, like me, but I'm not sure. He has constant swelling of his forehead, fingers, fevers of 102 at least once a month and pain daily everywhere, which increases with age. Do my son's symptoms sound familiar to anybody? Also, I have severe CFIDS, Gluten Intolerance, etc. Please pray for the answers to Kyle's issues be found out through your answers or the screenings. It is cheaper to pay Pediatrix the almost \$90, which equals 3 co-pays, and of course a lot cheaper than having a medically fragile child! I'm getting the, "your crazy" thing again, and tired of chasing after the wind, especially since I'm so sick. I pray that he will be o.k. until I figure it out. I wanted to write this story to give hope, and love to others. My faith in God, has pulled me through the darkest hours, loneliness and constant grief!! I persevere because of my faith and because of Hannah's dream to be a doctor one day to research other possible cures for GA1, and other diseases. She actually shared her dream with me without any prompting!!!!!! Hannah has taught me the value of life, patients, perseverance, and how to speak up etc. If her life's story will prevent this from happening to anyone else than I feel blessed. I really want to encourage us to fight for newborn screening! My precious Hannah's story and other "angels" hopefully will spark that. After all, GA1stinks! Ha, Ha! Hey, I was also wondering if anyone else has noticed a similarity with GA1 kids having curly hair, slightly indented noses, and advanced before their crisis? Where are any other triple screens positive? Are any other moms struggling with autoimmune diseases? Are any affected GA1 kids able to bend at the waste? Please call 303-452-7796 I would love to share, cry, and encourage all of you, as well as pray for you all. You can also e-mail me at galstinks@yahoo.com, or bradwanda@comcast.net, or through the OAA listserv. Please understand I'm just learning how to e-mail. Phone calls are the easiest for me. Thanks for reading "Hannah's story."

God bless,
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Thornton, CO 80241
303-452-7796
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Hanna and her brother, Kyle.

OAA To Open Online Forum Discussion Group

The Organic Acidemia Association listserv currently has over 200 members and is the preferred listserv with information for all organic acid disorders. Members can join the OAA NEWS listserv by signing up on OAA's homepage - <http://www.oanews.org>.

The OAA will be opening a new Member Forum based communication discussion group on our new website. There will be rooms for research, insurance information, adults, teens and disease-specific forums. Only members will be allowed to read/share contributions written for each specific forum. This new discussion group will be introduced with the new OAA News.org website by year-end.

Recently other OA-related Yahoo groups have sprung which cover other OA disorders. These Yahoo Groups are not supported by OAA, but families may feel it necessary to become members of both or all of these groups.



Cay Welch and her son Michael talk with Hannah at the recent GA1 Fundraiser in Lancaster, PA.

Simona Lucas, Propionic Acidemia, Age 9

This is the story of my granddaughter.

Simona Lucas, born July 1996, came into the world a month early. Almost immediately it was evident that she was not like the other premies – she didn't move much and didn't really cry. She also didn't want to eat. Although they said they'd keep her at the hospital until her actual due date, she was sent home after only 10 days. Simona was still inactive, quiet, didn't eat much, and periodically her eyes would roll back in her head. At her one-week check-up, her body core temperature was lower than normal and her activity level hadn't changed. She wasn't taking any formula, wasn't having regular stools, and was limp and lethargic. The doctor said that this was typical for a preemie.

Two days later she wasn't doing any better. She was nearly unresponsive, difficult to wake, and wouldn't eat. We took her into emergency, but they just wrapped her in warming blankets for an hour and sent us on our way. Routine lab work was normal and the doctors again said that this was normal – premies sleep more and can have a hard time regulating body temperature.

Later that evening I awoke to Simona's gasping. I went to her and found her choking on thick white mucous coming from her nose and mouth. I quickly wrapped her in a blanket, woke her mother, and we were on our way to emergency again. In the car, my daughter used the suction bulb to clear away the mucous. In emergency they ran labs again and came in

to say that Simona was a very sick little girl – but they didn't know why. Her labs had been fine just a few hours earlier. She was air-lifted to DeVos Children's Hospital in Grand Rapids (about 40 miles away).

Upon her arrival she was admitted to the pediatric critical care unit. For two days she fought for her life while the doctors tried to figure out what was wrong. Twice she required resuscitation. Finally, urine and blood samples were sent to Boston Children's Hospital where Dr. Kelly diagnosed Simona with PA. She required 3 blood transfusions to clear the ammonia from her system. She had surgery for a G-tube and Neisen wrap. After finding the right combination of formulas, vitamin supplements, and meds she stabilized. The medical team had us stay an extra week so that we could learn, hands-on, what her around-the-clock care would be. (Feeds every 2 hours, meds, hooking up the heart monitor, infant CPR, and information on PA.)

At the time of Simona's birth, my daughter had just turned 16 and Simona's father was 17 – each with their own problems and not prepared to give the level of care Simona would require. Before her discharge from DeVos, it was determined that I be appointed Simona's legal guardian.

The Social Worker at the hospital gave us information on resources that are available for children with special needs. One of the resources was the "Early On Program". They came out to our home 3 weeks after Simona's discharge from the hospital. They started her on physical therapy twice a week, and at 18 months of age she began occupational therapy twice a week. At 3 ½, she began speech therapy. Visiting Nurses were another resource we were able to access for the first month after returning home. They came to read the heart monitor, check vitals, and answer any questions we might have. Initially, there were weekly lab runs to Grand Rapids, but now we only have labs every six months.



Simona has come a long way since then. She still continues to amaze me. She attends school in an SXI classroom with a few hours of inclusion in regular education classes each week. Simona loves school and the bus rides. School evaluations place her in the

1½ - 2 year-old range – but she is much brighter than her scores convey. She understands a lot and has a great sense of humor! She is unable to communicate verbally, but does say a few words and continues to work on feeding independently. She loves music and likes to play "cooking" and dress-up with her hats. At home, she likes to crawl around the house. Simona is able to walk a few feet with her AFOs, a walker, and someone to assist in guiding the walker. Her hips move in and out of their sockets and as she grows, it is getting more difficult for her to walk. She's graduated from her big stroller to a wheelchair and is able to move herself a little on her own.

We have been very lucky, having very few hospitalizations since her initial diagnosis

9 years ago. Her medical team has done a tremendous job working to regulate medication and diet and in keeping her healthy. Simona gives us reason to celebrate — not just her daily accomplishments, but having her in our lives!

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Those two little guy's are my grandsons, Simona's younger brothers. The one on Simona's left is Shane 7 yrs and on her right is Lance 6 yrs .

OBITUARY

Kay Tanaka, M.D.

March 2, 1929 - August 21, 2005

The inborn errors of metabolism community lost a pioneer and great friend on August 21, 2005 when Dr. Kay Tanaka, M.D. died. Dr. Tanaka was born March 2, 1929 in Osaka, Japan. He received his Medical Degree (1956) and a Doctorate Degree of Medical Science (1962) from the University of Tokyo Faculty of Medicine and Graduate School of Medical Science, respectively. Since 1963, he has lived in the United States where he held faculty positions at Baylor College of Medicine, Massachusetts General Hospital and Harvard Medical School, before being recruited by Leon Rosenberg to the Department of Human Genetics at Yale University School of Medicine in 1973. He became Professor of Human Genetics in 1982. For more than 20 years he directed one of the most productive research laboratories in the field of inborn errors of metabolism, and at the same time founded and directed the Biochemical Disease Detection Laboratory, one of the first of its kind worldwide. He retired in 1994 and was recognized as Emeritus Professor of Genetics in 1995.

Dr. Tanaka will long be remembered as one of the greatest scientists ever to turn his attention to the study of inborn errors of metabolism. Of his many, critically important contributions to the field, two are particularly noteworthy. His discovery of isovaleric acidemia in the mid 1960's led to the recognition of a new group of inborn errors of metabolism, the organic acidemias. Subsequently the purification of the mitochondrial acyl-CoA dehydrogenases by his laboratory and his characterization of them as a family of distinct, evolutionarily conserved enzymes revolutionized our understanding of fatty acid oxidation and branched chain amino acid metabolism.

An early research interest in the chemistry of fatty acids was a key factor in the seminal recognition by Dr. Tanaka that the "sweaty feet odor" noticed in two sisters presenting with

recurrent episodes of ketoacidosis was related to isovaleric acid. The persistence and rigor of his scientific approach is best appreciated by considering the systematic progression of his studies of isovaleric acidemia from the identification of the biochemical phenotype in the form of an abnormal metabolite, requiring the first application of mass spectrometry to the identification of diagnostic metabolites in urine, to the development of an enzyme assay, purification of the protein, cloning of the cDNA, and structural characterization of the gene. He brilliantly demonstrated that a dedicated scientist could continually adapt new technologies to develop an ever more complex understanding regarding a given disease. Equally importantly, he exemplified the importance of the partnership between basic science and clinical medicine by applying his new-found knowledge to improve the diagnosis and management of patients.

Perhaps the most significant of Dr. Tanaka's many contributions to the field of inborn errors was the publication in 1985 by his laboratory of the recognition and characterization of three mitochondrial fatty acid oxidation enzymes, short- (SCAD), medium- (MCAD), and long-chain acyl-CoA (LCAD) dehydrogenases. For more than a decade afterward, his group continued to provide critical insight into the structure and function of these genes and proteins and their role in human disease. For his accomplishments in this field, he was awarded a prestigious MERIT award in 1987 by the National Institutes of Health.

Dr. Tanaka was a man of unmatched scientific vision and powerful intellectual vigor, consistently a pillar of integrity. He demanded much of himself, and applied his rigorous standards to his many trainees. Dr. Tanaka's mentorship served as a professional introduction to inborn errors to many still



Picture taken in the early 80's of Dr. Piero Rinaldo, Cate Vockley and Dr. Jerry Vockley with Dr. Tanaka.

active in the field around the world. Their continued efforts in basic science and clinical practice will serve to amplify his impact for many years to come. While his health precluded much active participation at scientific meetings in recent years, Dr. Tanaka remained intellectually active until his death, and his friends came to value their contacts with him for their ability to stimulate new ideas and directions. He will be sorely missed.

Dr. Tanaka is survived by his wife Tomoko, also a physician, his son Atau, a renowned composer and researcher of digital music systems at Sony Computer Science Laboratory Paris, his daughter Elly, who has followed her paternal footsteps in science and is currently a faculty member at the Max Planck Institute of Molecular Cell Biology and Genetics in Dresden, Germany, and three grandchildren.

Submitted by:

Piero Rinaldo, M.D., Ph.D.

Paul M. Coates, Ph.D.

Jerry Vockley, M.D., Ph.D.

Early this year, OAA opened a fund to specifically help support research activities at Dr. Jerry Vockley's lab at University of Pittsburgh Medical Center, Children's Hospital of Pittsburgh. Families are encouraged to start fundraising towards this fund. Donation monies can be send to:

OAA/IVA Research Fund
13210 35th Avenue North
Plymouth, MN 55441

The OAA would like to announce that the OAA/IVA Research Fund has officially been named the Dr. Kay Tanaka Isovaleric Research fund in his honor/memory. This fund will assist Dr. Jerry Vockley's research in the area of Isovaleric Acidemia.

Pregnancy and Isovaleric Acidemia

On September 29, 2005 at 5:41 am, Kent Robert Brooker joined our family. He has born a healthy baby weighing in at 8 lbs 11 ounces and measuring 22 inches in length. He has been such a happy baby and is so patient with his parents who admittedly only have some small idea of what to do with such a little infant. Within a week and a half of his birth however we received the results of his newborn screening and happily everything came back negative. He really is a nice happy and perfectly healthy baby and even the doctors agree! Now at this point you might be asking yourself what is an article about a healthy non-OAA baby doing in this newsletter? Let me go back.

My name is Elizabeth Brooker also sometimes known as Beth and I have Isovaleric Acidemia. Many of you might have heard of me and my story previously, but just in case you haven't, let me give you a brief summary. I was diagnosed when I was 8 after a couple of very serious hospitalizations, during which I experienced all the usual signs of acidosis. After my diagnosis I was put on a low protein diet, and formula, and so far my IVA has been pretty well in control since. In 2003, I was married and I graduated from Brigham Young University with a degree in Physics Teaching. Since then I have been teaching high school and enjoying our new home in Connecticut. Just this last month I delivered a healthy little boy who I am happy to say does not have IVA and this article is a bit about my experience during pregnancy and how it all worked out.

When my husband and I first got engaged, my mother insisted we go and speak with my metabolic doctor at the time and consult with him about having children in the future. Our visit with him was very encouraging as he assured us the chances that my husband was a carrier was like 1 in 500,000 and so our chances of having an affected child with IVA was slim to none. We were also told that previous to any pregnancy I should get more serious about my formula and carnitine intake (which I have always struggled with). Once we moved to Connecticut we were thinking more and more about starting a family so I made an appointment with the Genetics team at the University of Connecticut. They also expressed the same opinions as my previous doctor, so we were assured that it would be safe to proceed.

In January after being perfect with my formula for over three months we discovered that I was pregnant and we were excited! Immediately I made an appointment with an Obstetrician to consult with him and get him and his group on board with the IVA. Much to our delight the initial OB we met with had a nephew with PKU which although not essential to his understanding our situation, did help break the ice a little. I tried to provide him with all the literature my mom could find on IVA and pregnancy, which wasn't much, and we proceeded from there. He got in touch with the Genetic team and it was decided that this would be treated as a normal, not a high-risk, pregnancy. Meanwhile, all the things that go along with being pregnant started to happen to my body, including morning sickness. I have to admit that on a scale of 1 to 10, I really wasn't that sick, but I would probably put it at a 5. I struggled with just nausea at first, but then I started throwing up. The first thing to really unsettle my stomach was my formula. Now I've never really considered myself to mind the formula, I usually drink it super concentrated and unflavored just to get it over and done with. However, being pregnant it seemed to be the one thing that I detested (that and cheerios). There were some days that I joked I had tasted my formula 3 times that day, as opposed to the regular once like I was used to. The only remedy to this was that I just tried to take it in the afternoon or evening and avoided those early morning hours. I also found that on some days I just wouldn't have all of it. My husband commented once that I would get to a point where I was ok then I would force myself to finish the rest of the formula and it was when I did that, that I was unsuccessful at stomaching any of it. All in all morning sickness was easily managed with the usual taking hard candies and plain crackers to work, and thankfully after three months I was mostly cured of it.

Once I entered the second trimester, morning sickness subsided and I also regained a lot of my energy. I also got more tolerant of my formula, though I still had to be careful not to have it first thing in the morning. Also because my stomach could handle it, I increased my formula intake to meet the baby's growing needs. In addition, I got to increase my protein intake from normal foods. Instead of just having one serving of a high protein food a day I got to have two servings! I really grew to



enjoy my bagels with cream cheese, yogurt, ice cream, and grilled cheese sandwiches. The only other thing that pregnancy affected in regards to the IVA is that now instead of being wary of formula in the morning I had to be careful of having it at night, due to indigestion.

The second trimester flew by and next thing I know it was third trimester and I started thinking more seriously about the birthing plan. I met with my team of metabolic specialists (namely the nutritionist and the MD), and they put together a plan of action for when I went into labor and faxed it to the OB so they would have plenty of time to review it. Once again my protein intake was increased after blood tests indicated my leucine was low, and other blood levels were good. I was fortunate that all through the pregnancy I never became anemic, the formula provided well for my iron needs. The only other thing of significance during the third trimester was that due to my pregnancy peaking in the summer I suffered from extreme swelling. However, even though I was very swollen, I never did develop high blood pressure or toxemia.

On September 28, 2005 I went into the hospital to be induced (something that was not part of the birth plan, it just worked out that the baby was showing to be very large in the ultrasounds and my mother was in town and wanted to be sure and see the baby before she went home). The day before I went in for a non stress test and while I was there the nurses reviewed the protocol for the next day. As soon as I was admitted I was given an IV, one with the pitosin for the induction, and one for the D10 mixture with Carnitine in it. Every four hours they checked my ammonia and ketones and as far as metabolics are concerned labor went quite nicely. Only once did the ammonia

Pregnancy, continued on next page

OAA Establishes an OAA/PA Research Fund

This fund was established in August, 2005 to support research and better treatments for Propionic Acidemia being conducted in the lab of Dr. Toru Miyazaki at UT Southwestern. OAA is responsible for the distribution and accounting of these funds. A PA parent and OAA board member, Janice Boecker is the administrator responsible for the ongoing management of this fund. Donations and more information on PA can be found at <http://www.paresearch.org>

Kathy Stagni
Executive Director
Organic Acidemia Association

Pregnancy, continued from previous page

levels rise and that was quickly solved with a slight elevation in the amount of D10 given. In the end I am happy to say that even though I was in labor for 14 hours, then had an epidural, pushed for three hours, and ended 20 plus hours later with a Cesarean Section, the IVA played no part in the labor and delivery process and everything was great. After delivery I did have some trouble keeping down food for a bit as the anesthesia wore off, making the D10 IV crucial but once that passed, even the recovery went smoothly!

The first week after Kent's birth, I had to really cut back on protein so my body could handle all the reabsorbing that happens after childbirth. Five weeks later, I am now on a slightly elevated low protein diet to allow for additional nutrients needed for nursing. I feel great and as far as I'm concerned IVA is something that we had to be mindful of during this whole process but it certainly did not hinder anything or complicate things too much. Once again proving that even us weird OAA kids can have normal lives!!!

Elizabeth Brooker, 23 IVA
Mother of Kent Brooker (Unaffected)

Jenna Lynn Delima (age 7), Propionic Acidemia Lauren June Delima (age 4), Propionic Acidemia & Cardiomyopathy

The year 2004 was an especially bad year for hospital admission for both the girls. The smallest flu or virus would bring them in for at least 1 week (each). It's been 3 months since the last admission (and counting). Winter time will definitely be a challenge, but we're hoping they have developed some immunity from their past & frequent admissions.

We had our first family vacation this May! It was a mini 4-day Coastal cruise (Vancouver to Seattle to Victoria to Vancouver). We had so much fun and to think the only flaw was that they thought one of our baggage carrying our kids' machines and medical supplies posed a "threat" to cruise security. I quickly opened the baggage to the cruise officials and we were able to enjoy the rest of our cruise. Next year's vacation, I'm thinking we can go further...to Disneyland! I'm sure flying will pose another challenge, but we can't wait.

We had this false expectation that since Lauren was diagnosed prior to birth, she would develop more "normally" than Jenna. Well, that theory has been proven wrong in our case. Jenna suffered severe neurological damage from her metabolic crisis at 5 months, but Lauren's long-term medical instability (both from metabolic and common illnesses) shows to be just as damaging, if not more.

Jenna is now in grade 2. She's in an inclusion class and her "alternative" school fits her perfectly. Her class is comprised of Kindergarten and Grades 1 to 3 students all learning together. The multi-age framework of each class builds on the kids' mutual respect and leadership. Jenna has developed so much confidence both in and out of class that I think she'll become a performer! She especially likes imitating "Kimee" of Hi-5.

Lauren is in her second year of preschool. We have seen progress in her social skills and motor development. However, she still needs to work on her play with other students and verbal skills. We are happy to report that



Lauren was assessed and deemed NOT to meet the criteria for an Autism Spectrum Disorder although her hearing is still under evaluation. She is presently entirely tube-fed. Since she developed food aversion at age 2, we have stopped trying to feed her orally. We're actually seeing a great response in that her food aversion is now gone and she's actually putting food near her mouth to lick for a small taste.

The kids go for regular EEG & ECHO exams due to the close correlation Cardiomyopathy has with Propionic Acidemia. Their last exam in June, the Cardiologist decided to start Jenna on Metoprolol, a heart medication to slow the heart rate. Lauren has been on Metoprolol & Digoxin since she developed Cardiomyopathy in 2003. We try not to concern ourselves too much on Cardiomyopathy because we know our kids are being watched by the best Cardiologist in Canada. The Biochemical Diseases team at BC's Children's Hospital, on the other hand, takes our kids illnesses very seriously and responds very quickly to every small abnormality in their blood work. We appreciate their help and would again like to thank everyone involved in our kids' care for their tremendous work and support!

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What's Happening in OAA



2006 OAA Calendar

www.cafepress.com/organicacidemia



"PA Child meets the Queen of England"

Eilidh Duncan (PA, Age 4) meets her majesty the Queen of England at the Aberdeen Royal Children's hospital in Scotland this past September. Eilidh (pronounced aylay which is Scots Gaelic for the name Helen) is in the white sweater with her mom, Ruth Milne, standing behind her.



Congratulations to Lori Smith (PA, Age 30) and Robert Binder on their recent wedding held August 27, 2005 in Las Vegas, Nevada.



Dear Friends of OAA,
I would like to congratulate Jana Monaco and announce that she received the Directors Award by the HRSA/MCHB at their Partnership meeting on October 18th.

She, along with her husband, Tom and son, Stephen, attended the ceremony in Washington DC, where she had the opportunity to speak. She pointed out, (as she fought back the tears) that she is driven by what has happened to Stephen and that she is just one of many family members across the world working so hard for the same common goal of NBS.

Jana was nominated for this Award by the MCHB because of the broad perspective and work she has done with NBS and because of the work/education that she has done with her regional NBS collaborative. In addition, Jana has also been the catalyst for the LEND Program at Children's. Along with all of that, she has also managed to care for Stephen and run a household. WOW!

I think we all agree that this Award is well deserved and that Jana is a mentor for all of us. I'd just like you all to know about it. She's too humble to announce it herself, so I'm taking the liberty of doing it for her. CONGRATULATIONS!!!

OAA Revises Website and Mission

The Organic Acidemia Association is a volunteer non-profit organization whose mission is to empower families and health care professionals with knowledge in organic acidemia metabolic disorders. We support early intervention through expanded newborn screening, solicit contributions and distribute funding that supports research toward improved treatment and eventual cures in the areas of Organic Acid disorders.

Go to any page on the web site by clicking on the drop-down menu at the top of each page

Click here for the new online FORUM where you can find the information you want sorted by TOPIC

Watch for your OA kid here. A new picture appears from the OAA Kid Library each time you visit this page

Find the information you need on any OA disorder by clicking on it's name here

Meet our families! Click here for our photo gallery!

Go right to our family gallery by clicking here

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Organic Acidemia Association

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Organic Acidemia Metabolic Disorders

- ▶ 2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase Deficiency (HMG-CoA)
- ▶ 2-Methylbutyryl-CoA Dehydrogenase Deficiency (2MBCD)
- ▶ 3-hydroxy-3-methylcrotonyl-CoA lyase deficiency (HMG)
- ▶ 3-methylcrotonyl-CoA carboxylase deficiency (3-MCC)
- ▶ 3-Methylglutaconic acidemia or 3-Methylglutaconyl-CoA Hydratase Deficiency (MGA)
- ▶ 5-oxoprolinemia
- ▶ D-2 Hydroxyglutaric Acid (D2-HGA)
- ▶ Glutaryl CoA Dehydrogenase Deficiency Type I aka Glutaric Acidemia Type I (GA-I)
- ▶ Isobutyryl-CoA Dehydrogenase Deficiency (ICBD) 3-Hydroxyisobutyric aciduria
- ▶ Isovaleryl CoA Dehydrogenase Deficiency aka Isovaleric Acidemia (IVA)
- ▶ Malonyl-CoA Decarboxylase Deficiency aka Malonic Acidemia (MA)
- ▶ Methylmalonic Acidemia (MMA)
- ▶ Mitochondrial Acetoacetyl CoA Thiolase - (3-Ketothiolase) (BKT)
- ▶ Multiple carboxylase deficiency (MCD, holocarboxylase synthetase)
- ▶ Propionyl CoA Carboxylase Deficiency aka Propionic Acidemia (PA)

The world's leading source for Organic Acidemia metabolic disorder support and information.

The Organic Acidemia Association is a volunteer non-profit organization whose mission is to empower families and health care professionals with knowledge in organic acidemia metabolic disorders. We support early intervention through expanded newborn screening, solicit contributions and distribute funding that supports research toward improved treatment and eventual cures in the areas of Organic Acid disorders.

What are Organic Acidemias?

Organic acid disorders (OAs) are a group of rare inherited conditions. They are caused by enzymes that do not work properly. A number of enzymes are needed to process protein from the food we eat for use by the body. Problems with one or more of these enzymes can cause an organic acid disorder. People with organic acid disorders cannot break down protein properly. This causes harmful substances to build up in their blood and urine. These substances can affect health, growth, and learning. The symptoms and treatment vary between different organic acid disorders. They can also vary from person to person with the same organic acid disorder. [more>>](#)

Newborn Screening

Newborn Screening saves the lives of innocent babies each day. For more information and to find out if your state offers expanded newborn screening

In The News...

Fundraising Awareness Bracelets Are Here! >>

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Nutrition News

By Brittany Smith



There is a new low protein cookbook that is out, *Apples to Zucchini: A Collection of Favorite Low Protein Recipes* by Virginia Schuett and Dorothy Corry Available thru SHS 1-800-365-7354.

Enjoy Life Foods has several delicious low protein offerings. They produce soft baked cookies, snack bars, breads, bagels, and cereal. Their bagels and cereals are higher in protein, however. They are available at many grocery & health food stores, at some low protein vendors (www.lilsdietary.com & www.dietaryshoppe.com), and through their website at www.enjoylifefoods.com. Enjoy Life recently purchased Perky's Foods and will be coming out with a lower protein Cheerios-type cereal soon. All of their products are gluten-free.

Sugar Roasted Pumpkin *Cooking.com*

| | | |
|------|---------|-------------------------------|
| | 1 Small | Pumpkin, quartered & deseeded |
| 10 g | 2 Tsp | margarine |
| 37 g | 2 TBSP | Brown sugar |

Preheat oven to 425 degrees.

Place pumpkin wedges, cut sides up on to 9x13 greased baking pan. Spread 1/2 tsp margarine on each wedge, then sprinkle with 1 1/2 tsp sugar. Bake for 35 minutes or until tender.

YIELD- 4 servings. 1.5g protein and 100 calories per serving.

Soft Pumpkin Cookies *Adapted from Very Best Baking*

| | | | | | | | |
|------|-------|------|-----------------|------|---|------|-------------------|
| 310g | 2 1/2 | Cups | Flour | 244g | 1 | Cup | Pumpkin, canned |
| 4g | 1 | Tsp | Baking soda | 6g | 1 | TBSP | Egg replacer |
| 4g | 1 | Tsp | Baking powder | 4g | 1 | Tsp | Vanilla extract |
| 2g | 1 | Tsp | Ground cinnamon | 10g | 2 | Tsp | Water |
| 2g | 1 | Tsp | Ground nutmeg | 240g | 2 | Cups | Powdered sugar |
| | 1/2 | Tsp | Salt | 45g | 3 | TBSP | Water |
| 300g | 1 1/2 | Cups | Sugar | 15g | 1 | TBSP | Margarine, melted |
| 120g | 1/2 | cup | Margarine | 4g | 1 | Tsp | Vanilla extract |

Preheat oven to 350 degrees. Grease baking sheets.

Combine flour, baking soda, baking powder, cinnamon, nutmeg and salt in medium bowl. In large bowl beat sugar and 1/2 cup margarine together. Beat pumpkin, egg replacer, 2 tsp water, and 1 tsp vanilla extract into sugar/margarine mixture. Gradually, beat in flour mixture. Drop by rounded tablespoon onto prepared baking sheets.

Bake for 15-18 minutes. Remove from oven and cool.

Meanwhile, prepare glaze. Mix powdered sugar, 3 TSBP water, 1 TBSP margarine, and 1 tsp vanilla extract in small bowl until smooth.

Drizzle glaze over cooled cookies.

YIELD- 36 cookies. .97g protein and 118 calories per cookie.

Cornbread Dressing

| | | | |
|-------|-------|------|----------------------------------|
| 170 g | 6 | Oz | Stove Top CornBread Stuffing Mix |
| 45 g | 1/4 | Cup | Margarine |
| 56 g | 1/3 | Cup | Onion, raw chopped |
| 40 g | 1/3 | Cup | Celery, raw chopped |
| | 1 1/2 | cups | Water |

Boil water and margarine in medium saucepan. Stir in contents of package, onion and celery.

Remove from heat. Let stand 5 minutes. Fluff with fork.

YIELD- 6 servings. 3.1g protein and 165 calories per serving

YIELD- 10 servings. 1.9g protein and 207 calories per serving

The protein counts for the lower protein food items from Enjoy Life Foods are listed below.

Original Sandwich Bread 36g

0.8g protein & 80 calories

Snickerdoodles 28g

1.0g protein & 130 calories

Chewy Chocolate Chip Cookies 28g

.16g protein & 120 calories

No-Oats Oatmeal Cookies 28g

0.88g protein & 110 calories

Double Chocolate Brownie Cookies 28g

1.16g protein & 120 calories

Gingerbread Spice Cookies 28g

0.83g & 100 calories

Carmel Apple Snack Bar 28g

1.51g protein & 110 Calories

Coco Loco Snack Bar 28g

1.7g protein & 120 calories

Very Berry Snack Bar 28g

1.5g protein & 120 calories



We are always looking for new low protein recipe ideas - please email your suggestions to Brittany at brittanydague@yahoo.com or to OANews@aol.com

News from SHS North America October 2005

MySpecialDiet.com

SHS North America announces the launch of MySpecialDiet.com, a revolutionary metabolic diet management Web site dedicated to improving the lives of the metabolic community.

MySpecialDiet.com provides:

- Current and relevant **information and research on metabolics**
- A dynamic online **diet management tool and helpful tips** to help with **diet compliance**
- A **community** encouraging interaction among patients, families and healthcare professionals
- Best of all, membership is **FREE!**

Join this exciting online metabolic community by visiting **MySpecialDiet.com!**

SHS Becomes Exclusive Distributor of Milupa Products

SHS North America and Milupa North America are pleased to announce that effective November 1, 2005; **SHS North**

America will become the exclusive distributor of the Milupa range of metabolic products. SHS and Milupa have teamed up to provide the best in clinical nutrition to the benefit of the metabolic community. SHS North America now offers the most comprehensive family of metabolic products to help improve diet management and diet satisfaction. Discuss with your healthcare professional which SHS or Milupa product is right for you.

SHS North America will distribute the following Milupa products: MSUD 2 • PKU 2 • PKU 3 • HOM 2 • OS 2 • TYR 2 and UCD 2. *Please note that infant "1" formulations will be distributed until current inventories are depleted (expected in March 2006). Discuss with your dietitian or healthcare professional about transitioning to an SHS Analog infant formula or a Milupa stage 2 product.*

New and Improved Maxamaids and Maxamums

SHS North America announces the availability of its improved **Maxamaid and Maxamum** range of products. Features

include better tasting formulas and an updated vitamin and mineral profile, based on the new **Dietary Reference Intakes (DRIs)**, published by the Food and Nutrition Board of the Institute of Medicine. To ease adaptation to these new formulations, SHS North America is proving a sample pack along with a transition guide. Contact your dietitian or healthcare professional to discuss transitioning or switching to the new formulations. *Old formulations will be available for a limited time.*

Metabolic Minutes Newsletter

SHS North America has published the fall/winter edition of *Metabolic Minutes*, a free newsletter providing useful articles and information on metabolic nutrition. *Metabolic Minutes* also features a special patient corner and a list of community events. Contact SHS at 1-800-365-7354 to receive a current or past issue and to be added to the free *Metabolic Minutes* mailing list.



VitaFlo Announcement

Due to the growing demand for VitaFlo nutritional products in most of the USA, we would like to inform you of the establishment of VitaFlo USA LLC, a subsidiary of VitaFlo International dedicated to the sale and distribution of VitaFlo's products throughout North America. By early 2006 VitaFlo USA will be in a position to sell and deliver VitaFlo nutritional products to customers like you, as well as to help with your medical insurance payment coverage (when requested). During this

transition period, both Cambrooke Foods and VitaFlo are committed to ensuring that there be no interruption to the purchase and delivery of our products to you.

We are grateful for the support of our customer base in helping us to establish this USA sales and distribution network, and we look forward to beginning to serve your needs in the near future. We would like to take this opportunity to thank Cambrooke Foods for their outstanding customer service

on our behalf. Our main objective is to provide you with the same level of prompt, efficient service that you have received from Cambrooke Foods.

Mark Prizer
General Manager
VitaFlo USA LLC
888-VITAFLO (888-848-2356)
631-547-5984 direct telephone
631-367-2868 fax
mark.prizer@vitaflo.uk.co



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 Plymouth MN 55441

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www.oaanews.org

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Please make the following changes to my address/phone number/email address:

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___ I'm including \$5 for a family roster.

Mail to:

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

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