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Organic Acidemia Association

NEWSLETTER

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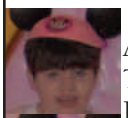
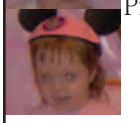
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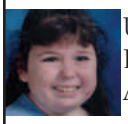
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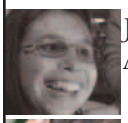
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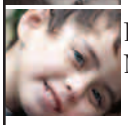
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OAA Family Group at conference in Dallas

Whew – we made through another conference year! I believe everyone who attended the conference felt it was well worth attending and everyone is excited about plans for the next one. The planning for this type of conference is an enormous amount of work, and I certainly could not have done it without Deb Lee Gould's help from the FOD Support Group. Eileen Shank, another FOD mom was also very instrumental in organizing the hotel arrangements for us – thanks Eileen! We had an equal number of FOD and OAA families in attendance. I want to acknowledge Dr. Charlie Roe from the Institute for Metabolic Disease and Baylor Healthcare for being our major sponsor to this conference. We certainly could not have done this conference without their professional/financial help, along with the other many sponsors (listed on page 9). We will need another major sponsor like Baylor in order to host another conference like this in 2008. If you have an interest in working on the plans for the next conference, please let me know.

The conference had its normal last minute changes – the first thing I heard after I arrived at the hotel in Dallas was that Dr. Regina Ensenauer from Germany was not going to be able to make the conference! Her airplane had mechanical difficulties in flight, and they had to turn around and head back to Germany. Regina was so disappointed, that she took it upon herself to contact the audio visual personnel at the hotel and helped to organize a 'web cast' of her presentation. I give lots of kudos to the staff at the Adam's Mark Hotel – they were so very organized in the audio visual, food, and staff – it was all handled very well and professionally. The only negative thing I can say about the hotel was that they were also hosting another conference with over 4,000 teenagers in attendance – whew, talk about a very busy hotel!

So after making agenda changes we were set to hear our speakers. Dr. Gibson, Dr. Roe started the morning with very informative sessions and then Regina's web cast went off without any problems right before lunch. I heard from MANY families how wonderful the lunch selection and the hotel staff were very accommodating with any special requests. The afternoon sessions went smooth … but as usual, we always went over schedule. Thanks to all the professional speakers and to the OAA board members, Jana Monaco, Kerri Wagner and Menta Pitre for helping to introduce the speakers. Thanks also to OA parents Rhonda Oberhelman and Joann Evans for introducing speakers as well. A big thanks to OAA board member, Jamie Pitre for video tapping the conference and I hope to have DVD copies available soon and will post information on the OAA listserv as to how you can obtain one.

There are many pictures from the conference available on OAA's new picture gallery on the OAA website. Check them out if you get a chance.

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.

Bjarki Gudmundsson, Isovaleric Acidemia, Age 20

Hello,

My name is Bjarki Gudmundsson. I was born on the 21.st of November, 1985. I live in a town called Selfoss, which is in Iceland. When I was about 2 years old I was diagnosed with a metabolic disorder, Isovaleric Acidaemia.

My father, Gudmundur Josefsson (1956), works as a carpenter. My mother, Elin Arndis Larusdottir(1956), works at the post office. I have two brothers, Larus Gudmundsson(1981) and Josef Geir Gudmundsson(1978). They both work as carpenters.

In the summer of 1987 I was taken very ill to a hospital. After many tests, the doctors discovered that I had Isovaleric Acidaemia.

Since it was discovered, I have always taken certain pills to help me break down the proteins. I take 5 pills Glycine and 1 vitamin in the morning, 5 pills Glycine and 1 tablet Carnitor at lunch and 5 pills Glycine and 1 tablet Carnitor at dinner. In addition I take 2 tablets of Calcium Santoz, which I dissolve in water.

I have many friends that know about my disease. The most common question, when I tell people about my disease, is "what do you eat at Christmas?" Many people would find it difficult, not being allowed to eat meat or fish. I have to say, that I'm getting used to it after all these years. I always find something to eat. There are some times when I can really feel that I have a disease. e.g. one time we went out to eat. We went to a steakhouse. When I looked at the menu I realized that there was nothing that I could eat.

Here is something that my mother wrote:

On 21. November 1985, a beautiful and healthy boy was born, named Bjarki Thor Gudmundsson. The first six months, he prospered well and was in good health, but at that time he nourished only on breast milk. At the age of 6 months he started having food with the breast milk, which he had until he was 13 months old. At that time, at the age of 6 months, he started getting sick and the illness increased until the second half of summer 1987. He was hospitalized at a hospital in Reykjavík very sick and almost unconscious. After several tests it appeared that he had Isovaleric Acidaemia. These times were very difficult for us the parents, his two brothers and the whole family. It became very difficult while we were adjusting to changed diet. Also the thought that the disease could have caused brain damage, but we knew that we would always love him despite what would happen. It later appeared that Bjarki Thor didn't have permanent brain damage, and since that he has performed well in school.

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Bjarki with his parents.

Amber, Age 10, and Tiffany, Age 6, Buice, Propionic Acidemia

Hi everyone!!!! First and foremost let me start by saying that I'm just a mom and not a writer, so please forgive me of any errors you may find, just remember this comes straight from my heart. Let me introduce you to the two most precious things in my life, Amber (PA) who is 10 and Tiffany (PA) who is 6. Our story began like most of yours did. Amber was born perfectly healthy, or so we thought, at 8 lbs, 6 ozs., while we were still in the hospital she started showing small signs of what was to come, she had trouble eating and starting sleeping a whole lot, the doctors reassured us that it was normal for a newborn to sleep that much and that she may just be having trouble breast feeding. We went home when she was four days old, and things changed that night. She would not wake up for feeding and she would not eat anything. Being new parents we were not sure if something was wrong or if we were just blessed with a baby that slept through the night. By morning I just had a gut feeling that something was not right and we took her to see her Pediatrician. Thank goodness for Dr. Gluckman, he was an experienced doctor who new immediately that something was not right and worked fast to get her to Children's Healthcare of Atlanta. While still in his office he did a spinal tap and sent us on to the hospital by ambulance, by time we reached the NICU at Egleston we had the results of the spinal tap, and knew that her ammonia level was 735 and that she was in critical condition. The next few days went by in a blur still with no diagnosis. We were in the best hands possible, our Geneticists, Dr. Paul Fernhoff and our Nutritionist, Dr. Rani Singh, were working around the clock trying to find out what was wrong, finally on the fifth day; we heard the term Propionic Acidemia for the first time. As you all know, it was overwhelming but also a relief to finally have a diagnosis. Amber was put on the right formula and medications, and within two weeks we were able to go home. Amber's first few years were very rocky, we were in and out of the hospital on a regular basis for acidosis and several "normal" illnesses but it seemed that when she turned two years old, someone flipped a switch and things settled down quite a bit. Medically, she has gone through several ups and downs since then but for the most part she has stabilized. She has had seizures, on and off through the years, and those are increasing as we get closer to puberty, but they are getting more under control each day, she has very low muscle tone through out her body and does need a wheelchair for long walks, she gets tired very easily and requires a considerable amount of rest each day, but overall she is doing better. Amber is moderately developmentally delayed. She is in a special education 4th grade class at a public school, and is doing great. This year she has met almost all of her goals and is very popular with all the kids at her school even though she has trouble with her speech. She has a g-tube, but eats all of her meals and snacks by mouth; we only use the g-tube for her formula and extra fluids everyday. Her favorite foods are rice and strawberries, this week. She is the sweetest, best behaved child we could have ever asked for. And then there is Tiffany, my wild child. We found out that she had PA when I was 36 weeks; the Geneticist did urine organic acids on me and found that I was spilling methycitrate. So when she was born by planned c-section, everyone was aware that she probably had PA also. When she was born she weighed in at 6 lbs. 13 ozs. and looked perfect too. We started challenging her system and within a few hours her ammonia level started rising, she was transported to Children's Healthcare of Atlanta and under the care of Dr. Fernhoff and Dr. Singh by the time she was 8 hours old. Her ammonia level only got to 350 and she missed the coma stage all together. We thought because of these factors she would be better developmentally and physically. That was not the case. She has actually been more unstable than her sister. In the past 6 years, Tiffany has been hospitalized more than 200 times for "normal" illnesses and high ammonia levels; she has had pneumonia, RSV, staph infections, and over 18 blood transfusions. She is totally g-tube dependent, her diet consists of baby foods, maybe one day soon we will be able to change that. She is also moderately developmentally delayed, and is in the same special education class as Amber, she is in Kindergarten and has also met all of her goals for this year. She also has a wheelchair for long walks but seems to have more energy than Amber. She is a happy child that never meets a stranger, and loves to be around other children. Their two personalities could not be more different, Amber is quiet and loves puzzles, and Tiffany is outgoing and loves anything that makes noise.

Amber and Tiffany both are on the following formulas: Propimex-1, Polycose, Prophree, Valine, Isoleucine, Carnitine, and biotin. Tiffany is also on buphenyl. We have been blessed over the years with the most amazing team of doctors, first and foremost is Dr. Rani Singh, our nutritionist at Emory, without her help and constant support we would have been hospitalized more often than we were, Dr. Paul Fernhoff who has never given up on us even when they nicknamed Tiffany, "IFFY TIFFY" because they were so confused by her.

I know that most of your stories are similar to ours and I would love to hear from you. May God bless you and your families and may it be a great time for our children medically.

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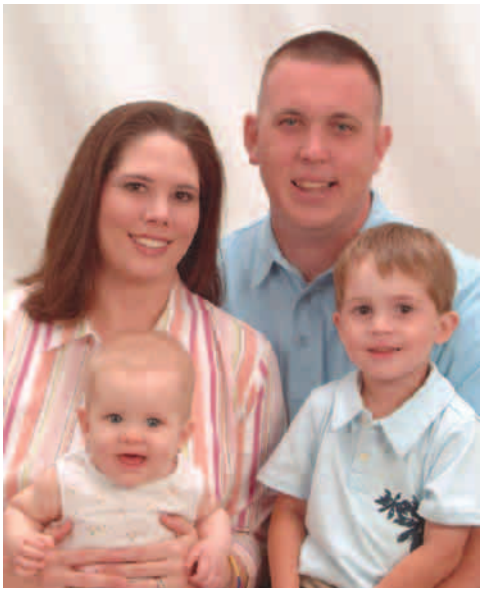
Cheyenne Posey, Glutaric Acidemia, Type 1, Age 10 Months



Hi Y'all! A little southern greeting to start our story!

My name is Lacy Posey. My husband, Shannon, and I have 2 children, Cody and Cheyenne. Cody was born on December 15, 2002. Although we are facing the "extremely picky eater" phase, he is a happy, healthy, rambunctious 3 year old boy unaffected by any metabolic disorders. Cheyenne was born on August 31, 2005. She has Glutaric Acidemia Type I (GA-1).

My pregnancy with Cheyenne was perfect. I gained 30 pounds, ate fairly healthy and stayed active. We opted out of the prenatal screenings because we didn't care if she had any type of disorders or syndromes; that wasn't going to change our minds about the pregnancy. No one in our families had ever exhibited any signs of genetic disorders before. Besides, no one ever thinks that it will happen to them. We weren't informed of exactly what tests are done on delivery or the option of having more done. I don't know that even with that information we would have done the screenings because of the lack of family history. We now realize how important it is to get the word out about the availability of these tests and how crucial they can be to your baby's health.

My delivery was induced because I was 2 days past my due date. When I arrived at the hospital at 6:30 am I had begun labor, so it was more of a help-along rather than an induction. The delivery lasted less than ? an inning (the TV was on ESPN). Cheyenne Grace was born at 1:03 pm weighing in at a healthy 7 pounds 11 ounces and 20.5 inches. She was so perfect! Our well baby stay was uneventful. We were released after 48 hours.

I took Cheyenne by my & my husband's offices on the way home. Then we went to our new house (we had only moved in 5 days before she was born) to see her brother and grandparents. About an hour after arriving home, Cheyenne was hungry. I got my new rocking chair, Boppie and blanket all ready in her new room to nurse. She wouldn't latch on right away so I waited a couple of seconds and tried again. The lights were off, so I couldn't see very well. Shannon came in to check on us and when he opened the door and the lights from the hallway landed on us, I could see that Cheyenne was blue. I handed her to Shannon immediately. He tried talking to her and moving her around to get her to breathe. When that didn't work, he began rescue breathing. I was on the phone with the paramedics. Cheyenne began breathing again, but soon had another apnea episode. When the paramedics arrived she was back to normal. Their devices are not made for 2 day old infants, so they suggested that we take her back to the hospital for a complete evaluation. We left for the hospital—thankfully only 10 minutes away. I want everyone to know that during all of this, my son was amazing. He wanted to know what was wrong with his sister and why "those men" were there. He has gone through just as much emotional strain as we have and he is an incredible big brother!

At the hospital, I asked for the pediatrician on call and the nurses immediately took Cheyenne to the NICU and us to a room. I was crying by this point. I had no idea what was going to be done. I thought we had just gone in to talk to the doctor and go home. I didn't realize the seriousness of the situation. The NICU RN asked us lots of questions about the events at home, pregnancy, delivery and well baby stay. I was so rattled! A few minutes later she came back in and asked to sign a consent form for all kinds of test. We agreed.

I went home to be with Cody while Shannon stayed at the hospital. I came back a couple of hours later. The first time that I got to see her, they had started an IV in her head and she was in an NICU bed under the lamp without anything on except a diaper. The doctors didn't want to take any chances of there being an infection before the test results came back, so they started antibiotics for a brain infection as well as anti-seizure medications. Her small veins had a difficult time tolerating the IV lines for more than 24 hours. Shannon and I stayed at the hospital continuously. They were gracious enough to let us stay in an unoccupied room only 2 doors down from the NICU. It didn't have a shower, but we would take turns going home to clean up and spend as much time as possible with Cody. Cheyenne had 5 more episodes during the first 24 hours in the NICU. After beginning the anti-seizure medication, they stopped. As for feeding, I continued to breastfeed the entire time she was in NICU. She seemed happy and healthy…especially compared to the infants around her. She was by far the biggest one in there! On day four the doctor ordered CT, MRI and EEG. The EEG was normal, but the CT and MRI showed bleeding and excessive fluid in her head. Thank God that we had a doctor that was educated enough to order a full battery of tests, including metabolic. I have heard so many of your stories about doctors that didn't even know a metabolic condition was in the realm of possibilities. We were released on September 8, 2005 after 6 days. Still no answers, but seizures were the best diagnosis and the nurse said that we would be receiving test results over the next 2 to 30 days.

On September 13, 2005 around 6 pm, we got the call. I knew it was strange for our pediatrician to call us and especially that late, but he had always been great so I just shook it off and answered the phone. He said that one of the tests had come back abnormal. He told me (of course my husband and son were out getting dinner!) that Cheyenne had Glutaric Acidemia Type I. He told me all about what he had researched, but urged that I not look it up until we see a specialist. I agreed. The only metabolic doctor in Alabama happens to practice near us. She called that night and scheduled us to come in for a 2 day admission to have more tests run, teach us about the disorder and start her diet. I thought "This is nothing…no meat, no problem! Wow, was I wrong! I don't think it ever really hit me until we got home and I began to read through all of the pamphlets and books we had been given. We had to switch to Similac (my breastfeeding was out the window…I cried), Glutarex-1 began being shipped and the talk of G-Tube surgery began. Our metabolic doctor still does not believe the apnea seizures are related to

Cheyenne's GA-1, so we are blessed to have found this disorder.

Cheyenne has only had 1 admission for metabolic issues. She was vomiting (not extensively), but our doctor wants to be overly cautious because she thinks that Cheyenne's condition was caught before any major damage was done. We had the G-Tube placed soon after this admission when she was only 6 weeks old. She eats so well by mouth. During one of our check ups, our doctor recommended physical therapy (PT) services to keep her on track. We are provided these through Early Intervention. Our PT has stated that Cheyenne's movements are erratic but not excessive. She also has been right on schedule with her cognitive, social and emotional skills, and her motor skills were only slightly delayed…but she has made up for that already!! She only comes out to our house once a month and it is mainly for monitoring purposes; she will come more often if the need arises. Since the initial PT visits, Cheyenne has not shown any more signs of a movement disorder…YEAH!!

Cheyenne began solid foods on schedule with infant cereal and now eats just about every vegetable, fruits and even spaghetti! She loves food! As for medication, she is on carnitine, riboflavin, zantac, pantothenic acid and co-enzyme Q10. She is allowed 6.5 grams of protein per day (soon to increase to about 10 when she is a year old). We only use her g-tube to feed her once a day for medicine and to keep her & us used to using it. Our doctors are so impressed that she eats so well by mouth! She weighs almost 21 pounds and is 31+ inches tall with big blue eyes and the blondest hair—like a little angel.

Overall Cheyenne's development has been typical. She smiles, laughs, plays and loves so much! She adores Cody and he is very protective of her. I believe that they are best friends. The dogs, Chloe & Chyna, are even in on the action and she loves to play with them. She just got her first pair of shoes and will be walking any day now! I am thankful that we do not have any major medical, financial or emotional difficulties. My husband and I both work full time. I work for the Shelby County School System in the Special Education Department and he is in the building industry. My parents, sister and in-laws take turns keeping the children during the week so we have a wonderful support system. We all wear the NBS bracelets. Every time someone asks, we are eager to tell our story. We are blessed to have Cheyenne's condition caught even without NBS and so early in life. With the tools to manage her disorder, we feel that we are already ahead of the game. As she continues to meet her milestones, we continue to be cautiously optimistic and enjoy every day we have with each other and our wonderful, beautiful children!

Thank you for listening and all of your feedback from the list serve. This support has been enlightening, heartfelt and appreciated!

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IOGA Social Event

When: Friday, September 15, 2006 at 5PM
Where: Clinic for Special Children
535 Bunker Hill Road
Strasburg, PA
RSVP: By September 7, 2006 to:
Cay Welch – 724-459-0179

Beverages and Meat Provided

Please bring a covered dish to pass

Dear Valued Supporter of the Organic Acidemia Association

OAA is very thankful for your continued and dedicated support. The most important resource we have is YOU!!! As it becomes increasing costly to further our cause, we have recently teamed with a company called SHOP4ZERO to introduce a revolutionary new way to raise funds to support and achieve our mission.

- Do you shop online?
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- In addition, you can tell all of your friends, family, co-workers, EVERYONE about your great SHOP4ZERO site and when they buy online, you and OAA will get a % of their Cash Back reward.

And so it goes, on and on, as others refer others to SHOP4ZERO, OAA will get a % of EACH person's Cash Back who buys online.

www.oaanews.shop4zero.org

Isovaleric Acidemia families at OAA/FOD Conference in Dallas

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Plymouth, MN 55441

Dr. Charlie Roe, Deb Lee Gould (FOD), Dr. Larry Sweetman and Kathy Stagni from the Dallas Conference

Update: Leah Masten, Propionic Acidemia, Age 8

Leah is now 8 years old. There have been many changes in the past few years. We moved to a new house, built an in-law apartment for her Meme who watches her after school, and inherited a new dog. (a black lab puppy). Leah also has 2 new cousins Rachel 1 ? years & Brayden 1 year. Both were born happy & healthy. She has been very healthy and is growing taller now. For a long time her weight was a major issue and her height had stalled. We installed an above ground pool for her last year and the exercise has been great for her. Also we found a therapeutic horseback riding place for her to learn to ride and strengthen her muscles. She loves it. Leah still enjoys doing puzzles, listening to music, and watching movies. She's a fairly easy going kid. She has a routine that we follow consistently that helps her to stay focused. She is in the second grade and is reading and learning nicely. She is a bit behind her pears both academically and socially but seems to be doing ok. She doesn't like school very much. She has been "tested" regularly since birth for one reason or another that she seems to be annoyed with school work at this point. The teachers try to make it interesting and exciting for her though. Not much gets her super excited. That has been our struggle this year. We found that when Leah wanted to"get out " of doing her work she was making herself sick and I would go pick her up. Finally we decided not to pick her up and she seems to have stopped doing this. We are going to have her tested for attention issues related to anxiety and stress to see if we can help her with this. Right now she is classified as "Other Health Impaired" because she does not fall into any other category which I think is hard for some teachers to grasp. She acts and does everything an 8 year old should do so why is she in special ed? And what affect does her Health Issue have on her learning ability? And will she get sick if we discipline her? I have tried to convey that she does have learning issues and just needs extra help and extra time to process things but it seems without a more definitive label some teachers don't "get it". And we even got a letter from her Docs stating that she can be and should be held accountable for her actions like any other child and it won't "get her sick". I am hoping this will be our most difficult year and from here on she gets the help and support she needs. It is tough sometimes for us and especially for Leah. We have started to see the pressure she feels she's under to do good and be good. Sometimes we just wish she could be a kid without all this other stuff. Feedings, Doctor appointments, Labs, special classes. So many restrictions. We just try to do whatever we can to assure she stays healthy and is happy.

We just had an annual check-up and now have a list of follow up appointments to do. She had an echo; she'll have an EEG and MRI in June and will be going back to follow-up with Dr.Korson's at Tuffs Medical Center in Boston. Leah's diet consists of her formula containing Duocal, XMTVI Maxamaid, Pediasure and Complete Amino Acid Mix. She takes Carnitine, Biotin, Dextromethorphan, Coenzyme Q10, Sodium Benzoate, Iron Supplement, B-1 & pyridoxine. She gets Zofran & Flagyl when needed and takes Zyrtec for allergy symptoms. She gets 3 feeding during the day with her Zevex, (the new Infinity) and an overnight. She has never been interested in eating but does enjoy chicken in a basket crackers and bugles' washed down with water. We have accepted that she may not eat and that is ok. We feel that if she is happy and healthy than that is all that matters. We love her the way she is and her happiness is the most important thing in the world to us. Thank you.

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Propionic Acidemia Research Network (PARnet) Begins Work to Advance PA Studies

June 2006 began a new chapter in promoting PA research with the formation of the **Propionic Acidemia Research Network (PARnet)**, an organization dedicated to finding and funding researchers and physicians who will join forces to develop better treatments and ultimately a cure for PA patients.

Over the past 30 years, numerous studies have evaluated various aspects of propionic acidemia. Unfortunately, little progress has been made to really help PA patients live longer, and more importantly, live better. Other than strict dietary control, caregivers have few resources to give their child a life that is not determined by the next feeding. Families and caregivers need better tools to deal with PA, and they need relief from the complicated daily routines often required to keep their children alive.

One obstacle to obtaining better treatments for PA is the distance between laboratory research and the development of useful medical applications. PARnet's mission is to bridge that gap, to create a network of researchers and medical professionals who will work together to develop new medical treatments that will truly provide PA patients a better quality of life. **By fostering communication between academic PA labs and the clinical community, our hope is to see medical applications evolve more quickly from basic science.**

A second obstacle that continues to hinder experimental data from being transformed into useful medical applications is the lack of funding for rare diseases like PA. To address this problem, **OAA has established the OAA/PA Research Fund** to provide grants to researchers who are looking for ways to improve treating PA. **PARnet endorses sending donations to this research fund, and emphasizes that 100 % of all contributions will go directly to research labs to provide money for salaries, equipment and supplies needed for a particular study.** Grant applications will be accepted annually and reviewed by the OAA Medical Advisory Board prior to the distribution of funds.

PARnet, in cooperation with the OAA is now accepting contributions to the OAA/PA Research Fund, to promote the work of highly qualified academic and medical researchers whose studies have the potential to make real differences for our children. You will receive a letter indicating your donation is tax-deductible in the U.S. when you send a contribution for the OAA/PA Research Fund to:

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jsboecker@paresearch.org

In the UK please send donations to:
Ruth Milne
Propionic Acidemia Research Network, UK
30 Morningside Avenue
Aberdeen AB107LX
Scotland, UK
oilfieldsales@aol.com

Checks marked clearly for the OAA/PA Research Fund may also be sent to:
Organic Acidemia Association
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Clinical and Scientific Studies on MMA and Cobalamin Disorders at NIH

Charles P. Venditti MD, PhD and Jennifer Sloan MS, PhD

June 2006 marked the second year anniversary of the NIH clinical and scientific programs. We are very grateful for the participation of thirty eight patients who have taught us new lessons about the clinical characteristics of MMA and cobalamin disorders. The studies continue to have open enrollment and we would be delighted to talk with any families or referring physicians about the clinical research program. We are particularly interested in meeting children and adults with MMA and organ transplants and 7 such patients have participated in our studies, including those with liver, kidney and combined liver-kidney transplants. These patients will help us gather information about MMA and transplantation and have provided insights into MMA metabolism that we have then studied in our mouse models of MMA. Additionally, one family whose child underwent transplantation donated their child's liver to our research study and this liver, which was otherwise unusable, became a critical reagent in gene therapy experiments we performed in the lab. Our clinical research interests also include the stroke syndromes and nervous system problems seen in some of the patients, the eye disease present in cobalamin C, the renal syndromes seen in MMA, and sibling adjustment issues.

In the lab, we have recently published articles on genetic and genomic approaches to study methylmalonic acidemia and have created worm models to examine MMA and cobalamin metabolism. We continue to study and create MMA mouse models and are using other organisms to examine the effects of malfunctioning of cobalamin metabolism. Our mouse models have allowed us to examine pathological mechanisms underlying the organ problems in MMA and have been useful in gene therapy studies as well. Some of our clinical and laboratory findings will be presented at the American Society for Human Genetics Meeting in New Orleans in October.

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Alya Wiseman, MMA, Mut 0, 3 years old

Our journey started when I was a few weeks from delivering my second daughter. My doctor started saying that the baby was measuring a little smaller than she should. Of course, her measurements were always borderline by the ultrasound so we did not plan for an induction. However, I was so worried about my baby having intra-uterine growth retardation that I went and read everything I shouldn't have from all of my books from nursing school. This of course, only made me worry more. Now looking back, I had much need to worry but for a different reason. A few days passed my due date, I was walking into work, which is also the labor and delivery unit, and wouldn't you know, that as soon as I open the doors to the unit, my water broke. Well, 26 hours later, on November 26, 2002, at 12:36 pm, Ayla Elizabeth Wiseman was born.

By a couple of hours after birth, she had a pretty significant sounding heart murmer that the pediatrician said was benign. It did go away by the next day. At about 18 hours after birth, breast-feeding started to not go so well. She would go on the breast like she was starving but then come off. She seemed frantically hungry but wouldn't stay latched. She had also started a slight grunt while exhaling that morning. I had the other nurses and even the pediatrician take a look but no one thought anything of it. At 24 hours of life, we were discharged from the hospital. With Thanksgiving being the next day, we were ready to get home and have some good times with the family and plenty of food.

Well, like with the rest of the stories, the breastfeeding got worse. She would act like she was starving but not latch on. We ended up giving her some formula with a syringe just to make sure she got some. Her grunting was a little worse so I held her in bed that night so I could keep a close watch on her. I awoke the next morning from a terrible dream where a friend of mine, who is a nursery nurse, looked over Ayla and told me to call 911 and get her to the hospital right away. Upon awaking, I noticed her grunting was much worse and she wouldn't eat. In fact she had started retracting a little bit, which is a sign of respiratory distress like the grunting. I called over a friend of mine in the neighborhood (also a nurse) and she agreed that we should take her in. She said her kids had the same symptoms with RSV as babies.

By the time we got to the clinic on Thanksgiving morning, at almost 48 hours of age for Ayla, her temperature was 94 degrees. Now she was getting pretty lethargic also. An ambulance took us to the hospital where she was put in the NICU. It was not long after that, maybe a few hours, when the doctor looking over Ayla told us he thought this was a metabolic disorder and that he had the metabolic doctor in with her at that time. The doctor said he had just read an article over this type of thing about a month before to refresh his memory, and while the symptoms were similar, he wasn't sure if that was the case. He was airing on the side of caution, and while he was having the metabolic doctor consult on Ayla, he was also treating her for any type of infection.

Dr Freedenberg, the geneticist, went over a million questions with us. We couldn't get a definite answer out of her or anyone for a while, which was so frustrating. The decision was to transfer Ayla to the Children's hospital where they had dialysis machines just in case she needed it for her high ammonia levels. They also had a sort of experimental medication there that they used for lowering ammonia.

Once at Children's, she was put into the PICU where she stayed most of the time. On December 3, we were finally so frustrated about not having anyone tell us for sure what Ayla had and what the prognosis would be that we threw a fit with the staff. Finally, an attending came in to talk with us but it was right after my husband had left for a breather. The doctor went ahead and told my mom and me that it is more than likely the worst type of MMA, which is mut 0, in which the kids live about 2-4 years of age. While we wouldn't know 100% for sure until the skin biopsy came back, they were pretty certain this was the case. I think hearing this and then having to tell my husband when he got back was the hardest thing I have ever done.

Luckily, my family and friends were always supportive and the metabolic doctor tried to reassure us that each case is different and that the information from the books is old. So, after three weeks of being in the hospital, we went home to try and figure out how to get back to living.

The first 9 months were hospital free. After she reached 9 months of age, 2 hospital stays a month began. We were in on her birthday, my birthday, got out just in time for Christmas and went back in before New Years. We made the decision to put in a port-a-cath because she was such a hard stick. Too many times she would decompensate in the ER just trying to start an I.V. After 3 bouts of blood infections with gram-negative bacteria (bad stuff!), we took that port out and put in another one and threw in a g-tube also. The g-tube has been wonderful and I can't remember trying to get along without it. The port, though risky with infection, has been a life-saver for Ayla I'm sure

We have had problems with growing but her human growth hormone level is actually high. This means that her body is trying to grow. It's likely due to her being acidotic much of the time. We have been able to attend NIH twice now and we are very grateful for everything we have learned. We are also very appreciative of any new updated information that Dr Venditti can provide us and other families. As of now, it looks like her kidneys are okay. We have not been able to do a 24 hour urine which would tell us more but her labs overall are okay. Her physical development is a little behind but with all of the hospital stays, I am not surprised. Her verbal is also a little behind but once again, I am not surprised. She still gets sick a lot but we can manage much of it at home. This is a good thing being that we did not have a metabolic doctor in Austin for quite a while. While we miss Dr Freedenberg, we are happy to have Dr Matalon who is doing a satellite clinic from Galveston Texas once a month starting this last June. He has been very persistent and aggressive at treating Ayla, which is what she needs. He has us do follow up labs and has the nurse call us regularly to check in on Ayla. We feel very well taken care of.

continued on page 9, Alya Wiseman

Jessica Osran, MMA- CBL c, Age 13

The teenage years have arrived. We have always told people that Jessica is developmentally delayed. Well, not when it comes to her recent physical development and teenage like attitude.

Jessica was diagnosed with CBL c at 6 weeks old at 4 lbs. 10 oz and has remained on her own growth curve. We now don't really have to add oil or Polycose to any of her foods as she now eats plenty. We monitor her protein at 20 grams per day.

Last summer Jessica attended the Illinois School for the Visually impaired for a Fine arts program, where she would call home every night, requesting to go to school there. It was quite upsetting as it is a residential school, five hours away, and we're not quite ready to let her leave home. We realize that through the good works of the school, many of the kids are awakened to the possibility that there are places of learning where, others who have visual impairments learn in a similar way. This is what Jessica came back to us with this notion that she wanted to go to a school with kids that are more like her. She realized that she might have options to her regular school, one on one aid, vision itinerants and learning disabled classrooms.

Fast forward to today, through Jessica's urging, and exhaustive research, I found a public school with a program for Visually Impaired children. She is in a program with other 6 other children reading and math had skills at a similar level. Her teacher is a teacher of the visually impaired (read not itinerant). She has an actual gym teacher who knows how to teach to the visually impaired and orientation and mobility teachers on staff, which she sees several times a week.

Jessica and I have started to learn Braille. Jessica wants to be able to read without all the eye fatigue related to concentrating on print.

We fell very well treated in our school and we are very happy for the current placement. She had a great year and is looking forward to 7th grade. It was worth all the hard work, and especially the concern of our school district for what is best for Jessica, to get her into a school program that she loves with children that accept her and invite her over for play dates and sleepovers.

Medically - we knock on wood lot don't we - Jessica has remained stable. After our very informative visit to the NIH last March, we increased her B 12 shot to daily, and decreased her cystadane to 9 grams two times a day. She remains on 1 gram of folic acid and 10 ml of carnitine 3 times a day. Our experience with Dr. Venditti and Jennifer Sloan was wonderful. Jessica is one of the older patients in the MMA study. Dr. Brooks the Ophthalmologist indicated that her eye's looked different than other's he has seen. The photographs of her eyes had to be retaken, as Jessica had a hard time holding still for all the bright flashes. Jessica accepted a bribe of a new pair of shoes to remain still. It's amazing what bribes can do.

Dan and I visited Dr. Rosenblatt in Montreal while we were at a graduation at McGill University. We had a great conversation about the gene identification, family testing, good restaurants in town and the future of B-12 research. Montreal is a beautiful city, very European in feel and luckily, most everyone speaks English.

We asked Dr.Rosenblatt if he would contact other Doctors who may have patients (children) with CBL-c as Jessica really wants to meet other kids that have the same disorder. If you are reading this and we have not talked about your child with CBL c please call us, especially if you have a daughter around Jessica's age.

We love to travel, call us and we can talk about maybe getting together. Jessica and her almost 10 year old brother don't fight too much in the car or any where else, so it makes traveling much easier.

We look forward to hearing from you. We wish all the best to the OAA families.

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Alya Wiseman, continued from page 8

We are very blessed in our journey to have met so many wonderful people. All of the people where I work, all of my neighbors, most of the people at Children's hospital, and now we can add all of the people we have been able to meet at NIH and at the OAA conference. Luckily, my family is very supportive and has been a much needed strength in our lives. I have seen the presence of a higher power more times than not and have experienced and felt things that couldn't be explained. Ayla is a true miracle in our lives and she and her older sister Hailey bring so much happiness and love into each day. I do not know what the future holds for us, but the one thing I know is, we will enjoy today for all it is worth, and for all we have in love and in life, which is more than I ever could have asked for.

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Kumiko Boscarino, MMA mut0, Age 4

Kumiko Elizabeth Ariella Menopi Boscarino was born September 12th 2001, with continuing coverage of "The day that changed America Forever" droning in the background. I was 41 years old and had already had one miscarriage, so we had an amniocentesis done as early as possible. If they had known to look for MMA they would have found it, but without any family history to indicate a problem they didn't think to look for such a rare disorder. My mother is Japanese and my father is of English blood from Virginia and Maine. My husband is a mix of Irish, English, Italian, and American Indian. It seemed that we had a pretty healthy mix of genes going. There is a little diabetes in his family and asthma in mine, but in general both our families are healthy.

Kumiko's two half-sisters were born when I was 19 and 21 years old, so any additional discomfort I felt during pregnancy, I attributed to my advanced age. This pregnancy did feel different, I had a great deal of pain in the lower right part of my abdomen, but every pregnancy is different. I also found that I couldn't eat meat from around the 5th month on, but this too can be pretty normal. Around the 7th month friends convinced me to really push soy, protein which I found easier to take, but still didn't enjoy (although I do now)

Kumiko is the smallest of all my babies and the longest delivery. It was all natural, no pain killers or episiotomy, and there were no complications during the delivery. Her APGAR was 9 and they told me babies born in Denver rarely rate the full 10 because of the thin air.

I breast fed my other babies, and in the hospital Kumiko seemed to take the breast just fine, but when we got home she refused to eat. We took her back to the doctor and she took a bottle just fine in the emergency room (although by then she was pretty hungry) so they told us to pump the breast milk and give it to her in a bottle.

During the next few weeks Kumiko was still not growing as well as expected so the doctor recommended adding formula to my breast milk. My husband was very happy with our new baby and 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. I know that every child is different, but her lack of interest in the world seemed very unnatural to me. The doctor was concerned that Kumiko was very floppy (hypotonic) but my other kids were also on the floppy side and my family has low blood pressure, so I thought that was just a family trait. Besides, her floppiness made her very cuddly and fun to hold.

By December, we'd seen the doctors quite a bit and she was still below weight, still not sitting up or holding her head up, and she seemed to spit up much more than other babies. We used mostly Enfamil, but also tried other formulas. When we tried formulas that contained "comfort proteins" she threw up and cried terribly. Again, we kept telling ourselves that every baby is different. I've heard of babies who screamed their whole first year, and mine was sweet and soft and gentle as could be. Still, she just didn't look happy.

One Friday afternoon in January of 2002, Kumiko really seemed to be getting worse and worse. She was really reluctant to eat and she threw up a great deal of every feeding. By Saturday afternoon we'd talked to the on-call nurse and thought she might have some sort of flu. We started giving her water and Pedialyte. She got to where she couldn't hold down any formula and just cried and cried. She didn't have a fever, actually, she felt a bit cold to the touch. Around 11pm Sunday, her breathing starting to get faster and faster until she was fully hyperventilating and we decided to bring her to the Emergency doctor.

Luckily, the hospital closest to us, just minutes away, was Denver's Children's Hospital. As soon as we got there they whisked us into a room and gave her oxygen. They suspected elevated ammonia in her blood and proved it with a blood test. The ammonia was making her blood acidic and was acting like anti-freeze keeping her cool too. They put her on a glucose drip with added bicarbonate to adjust her ph. They told us they suspected a metabolic condition.

Everyone at the Children's Hospital was so wonderful, they really helped us feel better right away, they kept us informed of what they were doing and we felt they were doing the right things. Kumiko looked terrible, but their monitors showed that they were getting her stabilized. By mid morning she had several IVs and was sleeping though everything.

They put her in ICU where she had a dedicated nurse monitoring her. Finally, I was so tired that I had to go home, but my husband stayed behind. It's so hard to leave a little one in that kind of condition, but we've found it's very important to get your sleep too. It's a difficult balance to achieve, and at that early stage we didn't know how many days off work we would need. We took turns working and staying with Kumiko and sleeping.

Within a few days they had a diagnosis for Kumiko's condition (MMA) and had a special metabolic formula for her. Kumiko woke up and her condition stabilized even more, to where we felt as though we were meeting our daughter for the first time. They moved her slowly from various IVs and medicines to the new formula. One medicine that continued and would become daily was Levocarnitine. As I understand it, Levocarnitine is a "carrier" substance that helps remove the Methamalonic acid from her blood.

We still use Levocarnitine, brand-name Carnitor, to help Kumiko clean her blood of the methamalonic acid. We never got a clear answer on whether this stuff is related to the L-carnitine that you can get at some health food or sports food stores, so if you know the answer please let us know. Sometimes Levo Carnitine gives your child a little bit of a fishy smell.

The doctors suggested that we have a gastro-intestinal feeding tube or "G-tube" installed in our baby so that in the future we can keep her hydrated even if she refuses to eat. Normally with G-tubes, a "nissin" is also performed for acid reflux. However, hey used some big machine downstairs and formula laced with radon to verify that her gastro-intestinal tract had no problems that would indicate that a Nissan was needed.

After nine days in the hospital we finally got to take Kumiko home. Kumiko was feeling some pain from the many IV sites and the g-tube 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. She began to make an effort to move more and slowly she began holding her head up.

During the months following her first hospital visit there were weekly blood tests, many doctor visits and prescriptions to pick-up.

We used Motrin more than Tylenol for Kumiko's pain, because the nurses explained

that it's easier on her liver. Since this condition is already hard on their livers, we tried to reduce other strains on it. I'm a big fan of aspirin, which is also metabolized through the kidneys, but it's hard to find it in a liquid form that you can give to babies.

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 go back and get another kind from a special pharmacy, and in the end she turned out to have a MMA type Mut 0 that doesn't respond to B12. On the other hand, they tell me that if your child responds to B12 it's really important to get it as early as possible.

I asked her pediatrician if there was something I could do about the pain of the shots. She recommended something called Emla or Lidocane to reduce the pain of the shots. Again, the pharmacy gave me the wrong thing at first, so it was a good thing I had the nurses check it the first time I used it. The Emla cream goes on the surface of the skin about 20 minutes before the shot, and then you have to cover it with a Telfa bandage to keep it from rubbing off (I used plastic wrap because it's less expensive).

The most important prescription was for the low-protein formula: Propimix from Ross. The Ross folks were nice enough to also give us a nice bag and mixing jug. I might seem small but during the stress of learning all these new things about our baby it did make us feel a little better to get that gift.

We were also very lucky that the State of Colorado had just past a law that required our medical insurance to cover special formula as a prescription, because the stuff is very expensive.

Kumiko at the park.

Another big adjustment was the g-tube. My husband and I both took the 20 minute class, but neither of us felt prepared at all. In theory we were supposed to feed her by tying a syringe above her crib and letting formula flow into her by gravity. In practice we found that once she was feeling better, it was impossible to keep her still and 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. We found that you have to go slowly and 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 of the consumables, so you have to learn to clean the tubes out and re-use them. This in itself is a big chore. We set up a hook over the sink for drying the washed tubes and a rotation schedule. We found that we could extend the life of a syringe by putting a little canola oil on the plunger after washing it. We found that you can get junk off the inside of a tube by pinching it from the outside so that the insides rub against each other. We learned to always keep at least one brand new syringe in her diaper bag in case one broke (our standard practice is to bring the last one we used, one clean used syringe and one set of new syringe and tube still in their packages. We also bring two wash clothes for catching the drips).

We were told that the g-tube sometimes leaks and we were given a box of gauze and tape each month. At first the g-tube site was a pretty weepy, but soon we found that the gauze seemed to hold the moisture near her and 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, when her second g-tube was about 5 months old we started having more and more trouble keeping the area clean enough. Kumiko, and who ever was holding her was continually adorned with a wet spot about the size of my hand. We had to change her outfits throughout the day, and she had an ongoing rash that we were treating with lots of gauze and diaper cream. Then one day, about six weeks after the rash started, the part of the g-tube that closes the hole when you're not using the g-tube, broke off. It happened on a Sunday morning when we had a lot of things going on, so I just made a temporary plug by taking a plastic tube tip, heating it and pressing the end closed. Well the new plug worked better than the old one and her rash cleared right up. It worked so well that she's still using it.

Today, Kumiko is five years old. Kumiko has been hospitalized three times, most recently in December of 2005 at Kaiser, her worst episode so far. At first, she was just vomiting and was admitted as a precaution. The next day after tolerating her formula, they released her because her tests indicated no elevated MMA or ammonia levels. 36 hours later, when we arrived at the emergency room again, Kumiko was lethargic, vomiting and dehydrated and definitely on her way to a crisis. Although we keep a copy of Kumiko's protocol letter in her "hospital bag", we were so panicked we forgot to check to make sure it was still in there. Instead, it was with her discharge papers from the previous day and there was a delay while they got hold of Kumiko's Metabolic Doctor. Too late- Kumiko went into a full blown crisis and needed transport to PICU at another Kaiser facility. She was there almost two weeks. Although she had been able to walk and was working on getting up stairs, the crises caused her to regress, and she has had to learn to do it all over again. She is a tough little girl, and works hard to make progress. She is just starting to work on stairs again, although she s still wobbly and weak when she walks. Her speech seems unaffected, and she is just starting to use two word phrases. She still likes to sing songs and listen to music.

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

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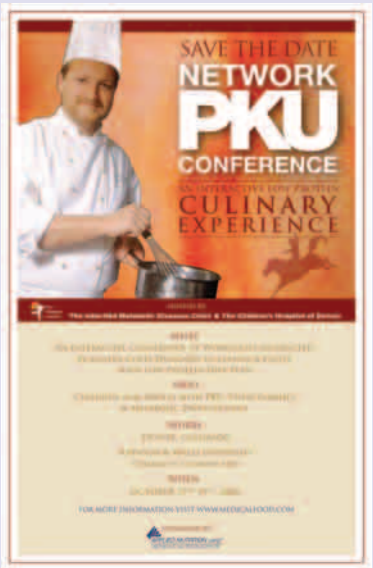

Eric Coldsmith, MMA, Mut 0, 17 years old

Eric Daniel Coldsmith was born September 20, 1988 in Chambersburg, PA. Just another boy who entered the world that day, but he was special, and going to be special in many different ways.

I was not going to have any children, possibly adopt I thought, but decided in my late twenties to have a baby. I was sooo excited getting the nursery ready, taking childbirth classes, living, eating and breathing, taking perfect care of my unborn child. No alcohol, plenty of rest, proper diet, I did not smoke and prayed every night for a healthy baby. The first red flag came when I had my last prenatal check up and the doctor said I need to prepare for a c-section….I said "C-Section!"?? You must be kidding; I have been practicing my breathing every week! I can look back now and laugh at how traumatic I thought that would be! I sat in the table and cried, but started to look forward to knowing exactly when I would give birth. I never dreamed I would plan it down to the minute.

The surgery went well, and at 8:00 PM, a 9-3/4 oz baby boy arrived with all of his fingers, toes and looked absolutely beautiful! I thought my prayers had been answered! I had no idea that was the beginning of a long road ahead…..

Eric got sick almost immediately, but it wasn't the hospital that even noticed. We saw him taking in a very little formula or mild, it seemed to be all running down his chin. We called for a doctor and he was put on an IV in the hospital a week after I went home. They sent him home thinking he was stable, but the poor feeding continued and development was very slow. Eric would spit-up once in awhile and cry once in awhile, but I thought he was a 'good baby'. When I went to the pediatrician, he told me to try some other formulas that some babies just didn't feed well, and it would maybe take some time to find a formula that would work for him. At 10 months of age, and all nutrition options were exhausted, the doctor ordered some blood and urine tests. He called us back and told us Johns Hopkins would be giving us a call because the results were a little far off from his experience.

I suddenly got very scared, and patiently waited by the phone and when he called, I will never forget those words "Mrs., Coldsmith, we do not want you to get into an automobile accident, but we want you to pack you and your baby's bags and get to the hospital as soon as possible!" We did just that! We spent 3 hours in the middle of the night talking to the doctors about some 'acid' thing that they knew a lot about, but didn't have all the answers for. They told us that special formula that was available, pending all the tests results. But what was the most devastating to me was the possible damage this high acid level can do to the brain and kidneys. If only someone would have thought of testing earlier! It was all coming together now; I had been feeding Eric everything he couldn't metabolize! I suddenly became ill and wished I would have visited more doctors and pushed to have something checked. He was taking enough in to maintain, but not to flourish; therefore, he never got into crisis mode. This is why he was so difficult to diagnose. We stayed at Hopkins for three weeks, and soon after Eric was put on his 'special formula'. I walked into the room and saw him actually sitting up in the crib! I didn't have to tell you how happy I was at that moment! After all the testing, he was actually diagnosed with "Methylmalonic Acidemia, Mut 0".

The years that followed were most difficult. All milestones were missed, of course, and some would never be obtained. We stated occupational and speech therapy immediately to try to catch up, but really never got too far. Soon Eric stated pre-school and went into special education. He is now 17 years 'young' and in the life skills class at the senior high in Shippensburg, PA, and only functions at a 1st grade level academically. However, it is still hard for Eric to understand why he is in the class he is in sometimes. He wants to be like the other 'cool boys' doing what they do, and going where they do, but they are all busy with their own lives, so Eric doesn't hand with a lot of friends. He was hospitalized a lot through those early years. Viruses, pneumonias, even common colds got the best of him. He had a hernia surgery as an infant and later on, a cataract implant. He also developed dystonia in both feet, which required three surgeries to straighten them and was also found to have "Legg-Perthes" disease, which required still another surgery to mend his hip. Needless to say, we knew the hospital very well, that is probably why Eric is till hooked on videos. Behavior issues also became apparent. It became so bad; he had to be admitted to the psychiatric ward for an evaluation. He has been diagnosed with anger problems and pervasive development disorder (PDD). He takes a small dosage of medications for that along with his MMA medications. We still follow-up with counseling. So depending on Eric's mood swings and how he is feeling, there are good days and there are bad days! Through it all, Eric is fairly stable now, and I would say 'slowly maturing' enough to understand why he will never have a driver's license, but can run his own go-cart. Because of his mental retardation, he doesn't understand the severity of his disease and would not eat a low protein diet and take care of himself if not watched very carefully. Thus, there has been a lot of 'food fights' over the years and it makes if difficult for him to go anywhere. I do keep him involved in swimming and moderate exercise, but also must see that his fluids are kept replenished from his slowly progressive kidney disease. He is on the Special Olympics swim team and does very well.

We still doctor at Johns Hopkins and visited Dr. Venditti once at the NIH last year. As you all know, there is no prognosis for this disease and no 'cure', only maintenance so 'everyday is a gift'! There is not a day that goes by that I don't worry for my son, but we try to live as normal a life as possible and I think positively. I could always be worse….Eric's creatinine is my guide.

But today is a 'good day'. Eric is well and is home. As for me, I have resolved myself into providing the very best care I can for Eric, to live as long and healthy life as possible. I continue to pray for him just as I did before he was born. We will continue to hope that Dr. Venditti will not only help the unborn and the newborns, but also find something that will help the lives of all the others.

I look forward to chatting with you all on the OAA listserv from time to time. It is nice to know that I am not alone.

Very sincerely yours,
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2006 Update: Health insurance coverage for OA medical-nutritional treatments

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Unfortunately obtaining and maintaining health insurance coverage of medically necessary monthly refills of; metabolic formulas, specialty low protein foods, medical supplies, and medications, continue to be a challenge for most families living with organic acidemias (OAs). Many of the suggestions originally published in the Jan 2003 OAA newsletter1 article continue to be applicable and hopefully still helpful to families obtaining or maintaining coverage by health insurance providers.

Virgina Schuett, MS, RD on her National PKU News PKU legislation and policies webpage2 (updated 1-06) has compiled data on 48 US states of which; 34 states have state laws (3 states listed as pending legislation), 11 states listed without state laws, and 24 states with state assistance programs. Of the 34 state laws listed, 6 state laws mandate coverage for people living with Phenylketonuria (PKU) only and 27 states laws mandate coverage for PKU and other metabolic disorders which may or may not include all OAs. There are often important differences in state laws and eligibility requirements for enrollment into state assistance programs. If you are a resident in a state with a state law then obtain a copy of your state's law for your files. If you do not currently have a copy, ask your metabolic clinic staff to provide you with a copy of your state law and/or contact your state's bill library to request a copy. If your state offers a state assistance program, ask your metabolic clinic staff if you/your child is eligible to enroll in your state's program.

Denials of coverage by health insurance providers seem inexplicable to metabolic clinic staff, even more so for families and people living with OAs, especially as medical and nutritional treatments are universally accepted as the current standard of medical care for the treatment of OA disorders.

WHY? Denials

When a denial is obtained, it is important to ask your health insurance to clarify exactly why the authorization request for coverage was denied. This information will best help you and your metabolism clinic and/or primary care provider to know what additional medical information should be provided for the appeals process with your health insurance provider. The reasons always seem to boil down to the basics:

1. Lack of knowledge/awareness about rare metabolic disorders and the medical necessity for uncommon and therefore expensive medical-nutritional treatments
2. Lack of consistent medical billing systems allowing for the appropriate billing and reimbursement to medical supply companies and pharmacies

Case example: Infant newly diagnosed with a metabolic disorder born in Massachusetts, metabolic clinic staff submitted a medical necessity letter and prescriptions for metabolic formulas to request approval of coverage by infants' health insurance provider. MA state law3 mandates coverage for patients diagnosed with inherited diseases of amino acids and organic acids by health insurance providers of metabolic formulas. MA state law also mandates additional coverage for specialty food products modified to be low protein limited to $2500 per year. Health insurance nurse case manager responds with approval of coverage of metabolic infant formula but parents told that formula coverage is limited to $2500/year. Parents informed that the infant's metabolic formula was interpreted to be the same as a low protein food and not a medical food. This $2500/year cap on formula coverage would only provide the infant with ~5-6 months of metabolic formula refills. Metabolic dietitian provided additional documentation that infant's medically necessary metabolic formula is actually a high protein formula, but metabolic disorder specific amino acid-free medical food as defined by the US FDA. "A medical food is prescribed by a physician when a patient has special nutrient needs in order to manage a disease or health condition, and the patient is under the physician's ongoing care. The label must clearly state that the product is limited to be used to manage a specific medical disorder or condition.'4 Infants' health insurance medical director reviewed additional information provided and approved metabolic formula coverage for infant as prescribed without an annual limit.

What? OA Spelling ABC's

If you are a resident in a state with a state law, always use the same terms written into your state law in conversations and correspondence with your health insurance customer service/nurse case managers. Always state the full name of your OA disorder and provide the correct spelling in your conversations. Not all state laws have the same terms and definitions which often adds to the confusion when referring to; metabolic disorders, metabolic formulas, and most of all the specialty low protein foods for health insurance providers. The table below provides a comparison of the terminology used in a sample of 7 different state laws; Connecticut (CT)5, Massachusetts (MA)3, Maine (ME)6, New Hampshire (NH)7, New Jersey (NJ)8, New York (NY)9, and Vermont (VT)10.

As long as your health insurance policy is not an ERISA exempt or self-funded or self-insured plan, then existing state law mandates should be applicable to your request for coverage as specified by your state law. Utilizing your state law's terminology should greatly help to reduce confusion on the part of your health insurance reviewers.

General term	State law terminology	State
OA disorders	'Inherited metabolic disease'	CT, NJ, VT
OA disorders	'Inherited diseases of amino acids and organic acids'	MA, NH, NY
OA disorders	'Inborn errors of metabolism'	ME
Metabolic formula	'medical food'	NJ, VT
Metabolic formula	'enteral formula'	MA, NH, NY
Metabolic formula	'metabolic formula'	ME
Metabolic formula	'amino acid modified preparation specialized formula'	CT
Low protein foods	'low protein modified food product'	CT, NJ, VT
Low protein foods	'food products modified to be low protein'	MA, NH
Low protein foods	'modified solid food products that are low protein'	NY
Low protein foods	'special modified low protein food products'	ME

If your state does not have a state law and/or your health insurance policy is an ERISA exempt plan, then continue to be consistent, politely persistent, and most of all patience, remembering that medical necessity is still on your side. Ask for a nurse case manager, and always note the names and phone numbers (extentions) of helpful and sympathetic contacts. Utilize your metabolic clinic's terminology whenever communicating with your health insurance customer service representatives. Ask your clinic

for a copy of the medical necessity prior authorization request letter so that you can emphasize from your personal perspective the medical benefits and necessity to obtaining health insurance coverage. Discuss and educate your health insurance about the risk of OA metabolic instability resulting in the potential for more frequent inpatient hospital admissions for acute medical management. Clarify the potential short and long term OA medical consequences to you/your child without the ongoing provision of medically necessary OA disorder specific treatments. Reference the updated May 2003 American Academy of Pediatrics policy statement that 'supports reimbursement for foods for special dietary use for inherited metabolic diseases and also calls for legislation to mandate consistent coverage for foods.'11

How? The Bottom Line…

In some cases, problems obtaining monthly refills of metabolic formulas, medical supplies, OA disorder specific medications, and low protein foods are not because of health insurance denials for approval, but because of difficulties with finding a contracted provider with your health insurance policy. Health insurance contracted providers typically include Pharmacies, Durable home durable medical equipment supply companies (DME), Home health infusion companies (HHI), and Low protein food companies (but not all low protein food companies offer health insurance billing programs). Which company provides your monthly refills depends upon the specifications of your health insurance policy. Your metabolic clinic or primary care provider would need to refer you to an approved contracted provider and write your OA disorder specific prescriptions and medical necessity letters. With a current health insurance prior authorization for approval of coverage, then your contracted provider(s) may start to provide you with monthly refills while submitting the bills on your behalf to your health insurance for reimbursement.

Some contracted providers have limitations determined by their company policies. Some contracted providers are not able to provide all brands of specific metabolic medical foods. Some companies are not able to assist patients who take their metabolic medical foods by mouth and will only provide refills to enteral tube fed patients, while other companies will provide monthly refills to both oral and tube fed patients. Most contracted providers are not able to provide monthly refills of specialty low protein foods.

Difficulties for DME/HHI/Pharmacies working with health insurance providers most often seem to be caused by problems with current medical billing systems and therefore the bottom line….adequate reimbursement rates equal to the actual cost of the products. Current medical billing systems include national Medicare, state Medicaid, and private payer (e.g., BCBS of MA, Tufts Health plan, Cigna, etc) medical billing codes. Formula companies submit their OA disorder specific medical foods to the US FDA for review to be assigned specific billing codes. But not all metabolic disorder specific medical foods are assigned billing codes that are recognized by all health insurers, and some codes are not defined to allow for adequate reimbursement rates to reflect the higher cost of developing and manufacturing metabolic disorder specific medical foods. Metabolic medical foods are much more expensive for formula companies to manufacture and develop compared with non-metabolic disorder specific infant, pediatric, or adult formulas. Metabolic disorder specific medical food codes should reflect this cost differential, but do not always seem to do so.

These challenges seem overwhelming. So what can parents and people living with OA metabolic disorders do to try to help clear up all this confusion?

OA 'To Do List'

1. Raising Awareness. Continue your efforts to raise awareness and educate others about OA disorders and medically necessary OA disorder specific medical-nutritional treatments. Try to gain advocates in all your communications with; your health insurance customer service staff, your health insurance case managers, and your pharmacy and/or DME/HHI companies.
2. Consistent Terminology. At all levels of the process, from the metabolic clinic's prescription and medical necessity letter to your communication with your health insurance and contracted providers. This should help to avoid miscommunication and prove the medical necessity of OA specific medical and nutritional therapies.
3. Organize Your Information. Obtaining and maintaining consistent health insurance approvals of coverage of medically necessary metabolic disorder specific prescriptions is like doing your income taxes. Gather contacts and work with your clinic by keeping track of deadlines for renewals of health insurance authorizations.
4. Utilize Resources. Every state has a division of insurance to assist health insurance consumers with concerns. In MA, the Office of Patient Protection provides all MA state residents health insurance information.
5. Plan Ahead. Allow for plenty of processing time, your health insurance prior approval process may take from 7 business days up to 30 days or more depending upon your health insurance policy, additional time is necessary for appeals of denials. Allow for additional time for your metabolic clinic and/or primary care providers to write medical necessity letters specific to your individual medical needs and communicate with your health insurance and contracted providers. Whenever possible, provide advance notice to your metabolic clinic and/or primary care providers with any health insurance changes and/or approval renewals requests. In our clinic, we typically ask for 4-6 weeks notice.
6. Shop Around, Ask Questions, Read the Fine Print. If you are given an annual choice of health insurance plans, ask in advance about benefits and coverage for your OA disorder specific medical needs. Remember to always check if your primary care provider, metabolic clinic and other specialty physicians are approved providers of any new health insurance plan.
7. Legislation and Advocacy. Contact and provide feedback to your state representatives if your state law isn't working for you, or if you think a state law would be helpful to your situation. We also need universally recognized and accepted medical billing codes for our metabolic disorder specific medical foods, medications, and low protein foods that are not limited by age, feeding route (oral or enteral tube), or form that also allow for appropriate reimbursement rates.

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