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Organic Acidemia Association

NEWSLETTER

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WHAT'S INSIDE

Keiko Ueda, Carolyn (PA), Mikayla (PA) and Melissa (PA) attending the NEPAD Conference in Natick, MA.

Dear OAA Families –

The OAA was founded on the belief that there is strength in numbers. Individually, the occurrence of each OA disorder is very rare. But collectively they represent a significant population. As a result, rather than establishing unique organizations for each OA disorder, we combined them under a single entity that could leverage the "clout" of the larger population. This single organization made it easier for medical professionals who could refer patients with any OA disorder to single point of contact for support and information. It also offered pharmaceutical and food companies a very economical way to reach the entire population of prospective customers for their products through a single point of contact and a single financial contribution. It also provided the basis for sharing medical research that could be used as a basis for improved treatment for all OA disorders.

Since its inception, the OAA has successfully executed its mission. This can be seen through our ...

- Tri-Annual Newsletter
- Newly designed Internet Website - http://www.oaanews.org
- OAA Listserv (http://www.oaanews.org/listserv.htm)
- Recently introduced Adult OA Listserv - (http://adultoas.oaanews.org/listinfo.cgi/adultoas-oaanews.org)
- Family Matching
- Bi-Annual Family Conference - this year scheduled in Dallas, Texas - June 23-24
- Research Funds for PA, MMA, and IVA

Over the years, the OAA has been the sole source for information and support for parents managing children who are afflicted with Propionic Acidemia. It has recently come to our attention that the Propionic Acidemia Foundation (PAF) has expanded its mission -- from raising money to support PA research -- to providing PA information and support services. We question the duplication of services that the OAA provides .... but welcome any additional information or services that help our PA parents improve the care of our PA Kids -- regardless of which organization provides it.

Please rest assured that the OAA will continue to serve the needs of its PA members as it has done for the past 15 years.

We have had some OAA members ask if we recommended joining the PAF. We are unable to assess the value that the PAF provides because the PAF has blocked the OAA from accessing its services. As a result, we are not able to make a recommendation on whether you should consider joining this organization.

If you do decide to join them, please remember that you will always be a valued part of the OAA family where you will continue to enjoy the strength that the combination of all OA disorders provides.

This issue of the newsletter has the registration form for our upcoming OAA/FOD National Metabolic Conference. We are very fortunate to have several excellent speakers. I hope that you will be able to join us in Dallas.

Thanks to the parents who contributed articles in this issue of the newsletter. Two families wrote about their child's experience while attending the MMA Study at the NIH (National Institutes of Health). You can learn more about the MMA study this summer at our conference – Dr. Chuck Venditti plans to be there to speak. Most interesting is the article by Lori Sanchez. Recently her son, Vincent had a liver and kidney transplant at Stanford in California. She is happy to report that he is making a wonderful recovery. Also we have published a press release from Dr. David Rosenblatt from McGill in Canada regarding his recent discovery of the CBL C gene. This is a major announcement and we thank Dr. Rosenblatt for keeping our families informed through the Organic Acidemia Association.

Melissa and I had the pleasure to attend the New England Connection for PKU & Allied Disorders family conference on March 11. The conference was held in Natick this year and as usual, they had a great list of speakers and offered an OA breakout session for our families. Thank you to Lynne Wolfe and Keiko Ueda who moderator our OA breakout session. We listened to Rani Singh from Emory give her update on Nutrition on Metabolic Disorders, and Dr. Harvey Levy is always so interesting to listen to as he gives us the history of newborn screening. Dr. Mardsen also gave us an update on her bone density study. Her study did not include any OA disorders, so there isn't much that I share with you on this at this time. Of course the best part of the conference is connecting with other families and visiting the vendor/low protein food booths. Melissa enjoyed meeting other Propionic Acidemia teenagers. The NECPAD plans their conference every other year – hope that if you live in the Northeast, you'll be able to attend some day.

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.

Melinda Bazzy, age 3
Robert, Isovaleric Acidemia, age 2
(photo taken 13 years ago)

On behalf of the citizens of the County of Wayne and the powers bestowed upon me, I am proclaiming this December 31st 2005 Melinda Bazzy Day. This proclamation recognizes your outstanding achievements, at such a young age, in helping to improve the lives of the many children afflicted with Autism in our County. For the past ten years, you have been the Chairperson of the Burger School Bowl-A-Thon designed to raise funds and build awareness of the many children afflicted with Autism in our County. Over the years, your efforts have made this event a tremendous success raising over $17,000 to support the students of Burger School. Furthermore, your wonderful articles about living with an Autistic brother have been an inspiration to many family members living with Autistic children.

You are a True Champion in helping to improve the lives of others in this County and I personally applaud your efforts. Burger School for Students with Autism also recognizes and applauds the impact you have made on the school.

You are a true example for all of the residents of Wayne County and an inspiration to the many young people who might not otherwise give their time to help our children with special needs.

December 31, 2005 is a Very Special Day in the County of Wayne, State of Michigan and the residents of Wayne County wish you a very Happy 16th Birthday.

Your efforts in helping your fellow citizens at such a young age are a model for all residents in the County. I wish you many years of continued success and I am very proud that you reside in our County.

Happy Birthday and thanks for improving the lives of many of your fellow Citizens.

Dec 15 2005
Date

Judge Maggie W. Drake
3rd Circuit Court
Wayne County, Michigan

Join OAA's New Adult OA Listserv

With the demand of older OA recently diagnosed, as well as our young OA's growing up - we have opened up a listserv specifically for Adult OA's -- Sign on by visiting OAA's website at **http://www.oaanews.org** or **http://adultoas.oaanews.org/listinfo.cgi/adultoas-oaanews.org**

Update: Hudson, Age 6-1/2 and Hope Wagner, Age 3-1/2 – Isovaleric Acidemia

The last time we wrote an article for OAA, our children were quite young and we still felt a bit inadequate in caring for their metabolic needs. A lot has changed in the past couple of years and I would like to share what's new with Hudson, age 6 1/2 and Hope age 3 1/2. Hope was diagnosed with IVA by NBS and Hudson was tested and was found to have IVA shortly after Hope's birth in 2002.

In 2004, our Metabolic Fellow, Dr. Regina Ensenauer, who worked under our Metabolic Physician at Mayo Clinic in Rochester, MN, was doing research on IVA. Because we knew Dr. Ensenauer and we were so interested in finding out just what mutation our children had, we joined Dr. Ensenauer and Dr. Vockley's study. It took a long time to get the results back, but we did eventually learn that our children suffer from a mild form of IVA, as our Metabolic physician had suspected all along. They both have a mild and a severe gene, known as the A282V mutation; which we have been told can be potentially asymptomatic in some IVA children. However, in our situation, we know that this is not necessarily the case, as Hudson struggled as an infant and toddler when his body would get stressed due to illness or well-baby shots. If you remember our story, you will know that Hudson was hospitalized several times before his diagnosis and he self-regulated his diet, refusing to eat high-protein foods, which he said gave him a headache (pre-diagnosis). Hudson always struggled to get over what others would refer to as a simple childhood cold or ear infection.

When the results of the study were revealed it really didn't change too much in our children's care. Hope remained on her formula, and both continued to be given a lot of latitude in their protein intake, due to their labs always coming back with low leucine levels. Hudson is currently not protein restricted, although he still self-restricts. He eats what he wants and his leucine levels always seem to remain in the neighborhood of 100. Hope is allotted about 45 grams of protein a day, but rarely takes that much in. Her leucine levels too stay in the area of 80-100. Because Hope has always been restricted, she is much more curious and will try foods that Hudson would never eat, such as shrimp or sausage, etc. Both children eat a high carbohydrate diet, and although we offer lots of fruits and vegetables, Hudson doesn't like to eat them either. He is a very picky eater but Hope is the best eater in the house, and the skinniest, I might add!

In August of 2005, Hope turned three years old; which was a big milestone for her. We had always struggled with her still taking her formula through a bottle, even though she would drink her juice or water from a cup. We talked at length with the team of therapists that worked with her and we'd tried everything we could think of to get her to drink it through a cup, to no avail. In August, we went back to the Mayo Clinic for the kids' check - ups. At that time, our Metabolic Physician, after collaborating with Dr. Ensenauer, and knowing how well Hudson did without formula, thought it would be wise to see how Hope fared without the supplement. We had always had a hard time getting Hope to eat and we thought it was due to the fact that she wasn't hungry, as the formula was doing what it is intended to do, provide the nutrients that IVA children do not get from table foods due to protein restriction. It was scary, but slowly, we started to wean Hope off the formula. It was a difficult transition for us, as most IVA's are on formula and we didn't have a lot of people to bounce ideas off of. It took approximately 4 months to wean her completely off, but she handled it like a trooper. What she missed most, was the bottle, not the formula. She has now been off formula for 1 1/2 years and she's doing great. She is on the small side, weighing only 28 lbs. and is about 37 inches tall.

Hudson is just the opposite of Hope. He tends to run over the 100th percentile for weight and height and at 6 1/2 years old is 57 lbs and 47 inches tall. Hudson is in his second year of AAU wrestling and although he does tire easily, we try to offer lots of fluids to keep him hydrated. He still has low muscle tone, but that is starting to resolve with exercise.

Together, with several other IVA families, we started an IVA support website, www.ivasupport.org. We have been contacted by many parents who wish to communicate with other IVA families and are looking for support and information about their children's disorder. For those of you on the OAA listserv, you will obviously note that there has been a lot of IVA babies diagnosed by NBS recently. If you have not already done so, I would encourage you to involve your children in Dr. Vockley and Dr. Ensenauer's IVA study. By doing so, you are contributing to research that will eventually help other IVA children and change lives. Dr. Ensenauer is the contributing metabolic physician for our website and she works closely with IVAsupport.org and OAA News to help families dealing with IVA. Dr. Ensenauer will also speak at our Metabolic Conference in Dallas, Texas June 23-24, 2006. I hope you can make it. I am excited to meet those of you that I have only connected with by email for the past 3 1/2 years.

In September of 2004, our physician nominated Hudson to receive a wish from the South Dakota Make-a-Wish foundation. His wish was to go to Disney World. Because his twin siblings, Hope and Haze, were still pretty little then, we opted to wait a year and a half to go on the trip. We just recently returned from Hudson's wish trip. I cannot begin to tell you how much fun Hudson had on the trip. He was treated like a King, as were his siblings, at Give Kids the World, where we stayed. If your Metabolic Child has not been nominated for a wish, I would highly encourage you to contact your physician to be nominated. As I understand it, parents may also nominate their children, but a physician must sign off on the paperwork, which states that the child suffers from a "life threatening" disorder. After all the blood draws, hospitalizations, formulas, tube feedings and worry that goes along with it, your child and your family deserve the wish. Please contact Make-a-Wish in your state to get information.

Lastly, I would like to thank everyone on OAA's listserv for always being so helpful and supportive when we have posted questions pertaining to IVA. It's so nice to have the expertise of all the other IVA parents. I have developed friendships with some IVA moms that I know will last a lifetime. I look forward to the day when my children can connect with yours and we can really share in the joy of FAMILY!!!

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Treatment of Isovaleric Acidemia

Jerry Vockley, M.D., Ph.D., Regina Ensenauer, M.D

Background:

Isovaleric acidemia (IVA) is an inborn error of the branched-chain amino acid leucine caused by a deficiency of the enzyme isovaleryl-CoA dehydrogenase (IVD). It was the first organic acidemia recognized in humans and can cause significant morbidity and mortality. It can present as a severe, potentially life-threatening disorder manifesting with acute neonatal coma in about half of affected individuals ("acute form"), and recurrent episodes of vomiting, lethargy, and varying degrees of developmental delay in the other half of patients ("chronic form"). Affected patients are at risk of episodes of acute acidosis and metabolic decompensation, usually due to intercurrent illnesses or other physiologic stress including fasting. During times of poor metabolic control the patient may present with the typical odor of isovaleric acid, also called "sweaty feet odor". More recently, a third group of individuals with mild metabolite abnormalities who can be asymptomatic have been identified through newborn screening of blood spots by tandem mass spectrometry. DNA studies of the IVD gene from patients with IVA have allowed characterization of different types of mutations in this gene. One gene mutation, 932C>T (substituting the amino acid valine for alanine at position 282 in the IVD protein), is particularly common in patients identified through newborn screening with mild metabolite elevations and who have remained asymptomatic to date. However, the consequences of this mutation regarding clinical outcome and the necessity of therapy are still unknown.

Treatment:

The majority of patients with IVA today are diagnosed pre-symptomatically through newborn screening by use of MS/MS which reveals elevations of the marker metabolite C5 acylcarnitine in dried blood spots. Isovalerylcarnitine in plasma and isovalerylglycine in urine are the hallmarks of this disorder, and are elevated regardless of a patient's metabolic condition. There are three aspects of therapy of IVA: 1) prevention of metabolic crisis; 2) dietary restriction of protein; 3) medication with carnitine (with or without glycine).

Prevention of metabolic crisis: Careful clinical observation of the patient regardless of the severity of IVA is required during times of metabolic stress (including illness and fasting). Sick day precautions for patients with IVA should include increased caloric intake in addition to decreased leucine intake. This is accomplished with oral solutions containing simple sugars and leucine free metabolic formulae or powders. Acute episodes of metabolic decompensation can present with vomiting, lethargy and signs of acidosis. Under these circumstances, immediate hospitalization is required so that IV access can be established and glucose administered. IV insulin may become necessary to maintain normal glucose levels in the blood. Reintroduction of oral intake including protein (0.5 gm/kg per day) with leucine should occur as soon as it can be tolerated; otherwise parenteral amino acids should be provided.

Dietary restriction of protein: If a child with IVA is on a protein-restricted diet, total protein and caloric intake must be adequate to support normal growth and thus monitoring of weight, length and head circumference is essential at follow up. In many cases, it may be sufficient to moderately reduce protein intake with natural foods to approximately 1.5 gm/kg per day. In patients with severe and recurrent clinical symptoms, leucine restriction in excess of total natural protein may also be necessary. The protein necessary to reach the recommended age-appropriate daily requirement must then be provided with leucine-free amino acids. Because leucine is important for protein build-up in the body, leucine restriction should not be too stringent to avoid potential side effects including muscle wasting.

Medication: The third aspect of therapy in patients with IVA is medication with carnitine with or without glycine in order to prevent toxic metabolites from accumulating in the body. Recognition of isovalerylglycine in urine in the initial patients with IVA first led to the use of glycine as mediation for this disorder. Isovaleryl-CoA is linked to glycine in a reaction that can be enhanced with extra glycine. Such supplementation prevents or lowers the accumulation of isovaleric acid in blood and reduces the length and severity of symptoms during febrile illnesses, but the optimum dose has not been determined. Patients show a dose sensitive increase in excretion of isovaleryglycine, but at least in one report an increase in the glycine dose from 300 to 600 mg/kg of body weight led to a decrease in the excretion of isovalerylglycine. Therefore, initial dosing in the range of 150 to 250 mg/kg per day is reasonable in patients with severe IVA. No reports of glycine toxicity have been published.

The identification of isovalerylcarnitine in blood and urine along with the frequent observation of a reduction in free carnitine levels in patients with IVA has prompted treatment with carnitine. A dose of 100 mg/kg body weight per day has generally been suggested and has been shown to increase the excretion of isovalerylcarnitine in urine. Combined therapy with carnitine and glycine has been shown to maximize the total excretion of isovaleryl-CoA conjugates, but the relative merits of the two therapies either singly or together in patients with more severe presentations including recurrent crises remains a matter of debate.

The optimal treatment for individuals diagnosed by newborn screening and carrying the common 932C>T (A282V) mutation is unclear at this point in time. Specifically, the potential for metabolic decompensation remains to be elucidated. It is important to observe affected individuals clinically, particularly when exposed to metabolic stressors such as febrile illnesses or fasting (e.g. when undergoing surgery). Additionally, low-dose carnitine supplementation (approximately 50 mg/kg body weight per day) is recommended if the plasma free carnitine concentration is reduced.

A better understanding of the variability of IVA and the clinical management of patients are among the challenges remaining in the study of this disorder in the upcoming years.

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Michael Dalton, MMA Cbl C, Age 17

MMA Families Participate in MMA Study at the National Institutes of Health

Our visit to the National Institute of Health. October 17 2005

Michael with his therapy dog at the NIH.

This fall we had the pleasure to be involved in a study at the National Institute of Health on MMA. Dr Venditti is very passionate about researching Michael's disorder. We feel very fortunate to have him involved in the research of this very complex disorder. Our days were pretty much filled with appointments. Dr. Venditti's team is extremely organized and helpful. Everything was very well planned. When we arrived at the airport we had someone meeting us to bring us to the Children's Inn. This is a place where families can stay while participating in this medical study. The Inn is right across the street from the NIH. We had a very nice room overlooking the beautiful grounds. The first night we were at the Inn dinner was provided for us. We felt very welcomed. There were many other families with different conditions, but all in the same situation.

Michael saw a number of specialists while at the NIH. He had a complete physical evaluation the first day. Dr. Venditti asked many questions! Some questions were hard to answer. If any of you are planning to be a part of this study, remember to write down all of your child's milestones and medical history. He also had a genetics evaluation done by Jennifer Sloan. She was also great! Michael fell in love with her. She is very knowledgeable about the metabolic pathway involved with the Cobalamin C disorder. The next day we had blood work done, pictures, x-rays and saw an Ophthalmologist. We did find out from the Ophthalmologist that Michael does not seem to have the damage to his eyes that is seen in many of the other Cobalamin C patients. He seemed to be puzzled by this. We have felt that Michael's vision has improved since he was a baby. He does not seem to cross his eyes as much and the strabismus is not as bad as previous years.

This was a day full of appointments, but Michael did great! He loves people and loved all the attention. On day 3 we had a developmental assessment done. I guess there were no surprises here. Michael is very delayed and seemed to be right on the track where we thought he was. He also was seen by a dentist. This was quite interesting. They had a machine that took pictures of his mouth/jaw in 3-D. On day 4 Michael was supposed to have a DEXA scan done, but we canceled this as he would not be able to sit still. This is a bone scan. We did have some x-rays done of his hands. This showed that he had a mild form of Osteopenia. Michael was also seen by a Neurologist. Dr. Venditti did feel that Michael's Homocysteine was too high at 99. He has been on daily injections of B-12 since we have come back from NIH. So far his level has not changed significantly. We are hoping that this will come down. I learned that high Homocysteine levels are the huge problem for the kids with Cobalamin C. Very interesting as I always thought it was the Methylmalonic acid that was the main problem.

Thank you Dr. Venditti and your team for doing this research. Hopefully this will help future parents and physicians understand this disorder. We are honored to be a part of your study.

Karen & John Dalton
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Sydney Adelaide Davies

(Methylmalonic Acidemia, Mut 0)
August 28, 2002-December 28, 2005

Bless This Family With Eternal Grace

Lord, be with these parents and ease their suffering
Their child was their ultimate offering
Let them find comfort in Your arms
Knowing their child faces no more harm
Losing a child seems so wrong
Help them move on now that she is gone
Their angel has gone home at last
Never to feel the pains of past
The parental love will forever be
Breaking the boundaries of eternity
An emptiness is left behind
This family needs You during this time
Help them to grieve the child they love
Help them find comfort in her eternity above
Sydney forever will be in their hearts
This child knows they are only a life apart
For there will come a glorious day
This family will reunite in Your name
In Your home of eternal splendor
The pain felt now, never again remembered
Ease this family's pain, holding them in your hand
Until such time as You decide they leave this land
Once again to be with their daughter dear
And again to their heart, hold her near
From this world was taken an angelic face
Now Lord, hold this family in your eternal grace

Johnny Tate, MMA Mut O

This is an account of the week we spent at NIH, participating in the clinical study for MMA Research. We encourage all parents of children with MMA to consider taking part in this study. All expenses for this clinical study are covered, including the travel to/from, lodging, food, and all of the testing.

Day 1 – Monday - As we drove into the NIH campus, we were in awe. It was like a small city. After we were cleared at the security check point, we proceeded to the Children's Inn.

The Children's Inn - after checking in, we received a tour of the facility. What a place! They have multiple game rooms: pool tables, toddler play rooms, rooms with internet access and computer games, arts and crafts rooms, and TV's everywhere. They have multiple large kitchen areas, a place to store medical supplies, dry food and refrigerated food, and private rooms with 2 double beds, a TV, and bathroom. They even allow free long distance calls at designated areas outside of your room. The staff is made up of full time employees and some volunteers. Everyone is extremely nice. There is local transportation to all sorts of places – food stores, shops, movies, dinner. Overall, it has the feel of a ski lodge, especially with snow on the ground.

Day 1 - Monday Night - We really just hung out and enjoyed the Inn. Johnny was happy to be there – we prepped him for the visit so he wouldn't be anxious.

Day 2 - Tuesday Morning - We could walk across the street or hop on the bus and ride across to the Clinical Center. The bus was sitting there waiting, so we accepted a lift over.

The new Clinical Center was complete as of 2004. It is a beautiful place. It has a Starbucks in the seven story lobby and all of the appointments are close by. We heard that what the pentagon is to defense the Clinical Center is to U.S. medicine. Very Impressive! See www.cc.nih.gov for a virtual tour of the center and the Children's Inn. We checked into admissions and then proceeded to the clinic floor and our room. The hospital rooms are semiprivate. Each bed has a plasma TV on this huge arm that allows kids to position anywhere. The plasma TV is also a computer, with internet and DVD/video game capabilities. We brought our game cube and portable DVD player, so Johnny was plenty occupied. There's also a nice playroom on the floor, with a lot of cool toys, books, and PlayStation2+games to play. They have therapy dogs, social workers, counselors, and plenty of friendly parents, children and staff. The Clinical Center is not an acute care, but a research hospital so it was really very quiet with only a few people on the patient floor.

Near the admissions area, there is a larger playroom with a sand table, tons of toys, and even guinea pigs. Since they offer to baby sit, we were able to talk to the doctors without Johnny tugging at us.

After checking in to the clinic floor, and getting acclimatized with our surroundings, we proceeded to the get the IV placed. It takes place on the ultrasound suite, since they offer an ultrasound guided IV. Johnny did well and did not need the ultrasound placement. A little lidocaine and the IV was in. He made all of the nurses cheer "Hip, Hip, Hooray", three times, because he was very brave and didn't cry. It was pretty funny.

The rest of the day was pretty light – one blood draw from the IV, some blood pressure checks, and just playtime for Johnny as we were interviewed by the doctors for a complete history. Your child and one parent stays at the clinic for the first night, and the other parent can return to the Inn.

Day 3 – Wednesday Morning - At 5 a.m., they come in and do what's called a Metabolic Cart – it measures the amount of oxygen intake and the amount of CO2 exhaled. It's pretty cool. If you are lucky, and we were, the little ones never even wake up. It's also referred to as a resting energy expenditure test to check the efficiency of his metabolism.

The rest of day 3 is pretty busy, but not crazy. We started with a morning blood draw from the IV, a two hour dental visit and then lunch. After lunch, we had an ultrasound of the abdomen to check his kidneys, pancreas and gall bladder. We snuck in a Hand x-ray (Optional) to measure bone density, and then pictures of the child and family. We rounded out the after noon with a meeting with the genetic counselors. Johnny was able to play in between visits, watch TV, play some video games, and cruise around in clinic in one of those toddler/child cruiser cars. Johnny and I would ride the elevator to the top floor, and stop in the chapel to say a few prayers for those in need. We took the portable DVD everywhere, just in case. It really helped when getting the abdominal ultrasound.

On Wednesday night, we were all allowed to go back and sleep at the Inn, but it is possible that, depending on the reliability of the test results, your child may need to stay at the clinic for another night. His levels were consistent and his IV became dysfunctional (from being so active) so rather than reinsert a new one, Dr. Venditti let

Johnny with Dr. Venditti at the NIH.

us go provided we continued with the urine collection for round 2 of the creat clearance test. Happy to be back where the action is, Johnny did some crafts and played more games. We ordered delivery from a local restaurant – Bethesda and the Washington area has plenty of good ethnic and American food establishments. We choose a Mediterranean place and really enjoyed it.

Day 4 – Thursday morning – bus ride back across the street to the Inn. We had a quick visit by the genetic fellows and staff – some 13 people came in the room for about 10 minutes, and then off to a meeting with the Developmental specialists. Johnny was great – he told Mom and Dad to wait outside while he tried to answer all of the questions. Afterwards, we had a meeting with the Nutritionists, and then an appointment with Neurology, which was held right in our room. We squeezed in a Dexa scan (optional) and added a visit with rehab and an OT evaluation to the schedule.

Day 5 – Friday morning – We had a long visit with the rehab doc, and received pattybobs (shoe inserts) and pencil grips, and books to help him learn to write from OT. I was surprised they provided a service like this but since families make the effort to come to the center, they explained there is an unspoken "social contract" whereby they spend lots of time with us in return for our bringing Johnny for testing.

The clinic stay concluded in the afternoon with a wrap up session with the whole team. It was very thorough. We reviewed the test results and discussed next steps and a variety of other topics of interest to both parents and doctors.

A bit more about the Children's Inn

They had really large kitchens, very clean, fully equipped, and brand new. A basket of baked

goodies was set out every day for guests, ample refrigerator space and your own locked pantry cabinet. They had other fridge space; one just for meds and another for specimen collections. Next was a neat library with a kid's section and plenty of recent magazines for us parents, a business center with internet computers and copy machine available all hours, a teen center with a pool table, a kid internet room, and lots of cozy places with TV's too. There's a free laundry room, also. Almost like a school art room, their crafts room had lots of art supplies, all free. As through out, the room was always open. I went in to the arts room during wee hours to grab some sparkly ribbon to tie around a dollar from the tooth fairy for Johnny. He lost a tooth that first night! We also had a mail box. We were only there a week, but received little gifts in our box every day from volunteers.

What John and I especially loved about the Inn and the Clinical Center is that everyone is screened before they come to make sure they aren't sick. They explained to us that bone marrow transplant patients and medically fragile patients stay there so infection control is very important. As Johnny played in the various play areas we felt so much better knowing that.

Since the Inn is across the street, we felt comfortable walking back to the room as one of us stayed with Johnny. We'd go back to shower, reheat our take out for lunch, make calls. Grab something for Johnny. Not anything like a hospital stay far from home (and a hot shower).

The Clinic Visit and some results

We were blessed with incredibly good interviews with genetics, nutrition, and Dr. Venditti. We learned so much as we had the luxury of asking many questions. There was no hurry, and they were all there, all ears, to listen to our concerns. After Dr. Venditti's explanations though I so wished I had taken biochem in college! Nonetheless, it helped that we sent him Johnny's previous labs so we could point out values of concern. We learned about things to work on with Johnny so he can succeed in school and gain strength where he is weak. Johnny gets OT/PT but its way too light to be effective as it turns out. We also explored why Johnny spills ketones, even though his diet has given him a BMI of 95%. The labs taken during our stay were ready and we received copies of all tests. Really nice to get real time results! With great care, we went through each finding.

For the first time, Dr. Venditti, and neurology, felt Johnny did suffer some neurological damage (at birth?). We will follow up on that finding with an MRI to be done at the NIH next year. His creat clearance was low normal, but needs to be watched over time. We also learned his pancreas and gall bladder were not quite normal, but otherwise ok. We discovered too that a few micronutrients in his diet exceed tolerable limits, yet we were giving him supplements on those. It reminded me that what made sense to do last year, doesn't apply now so we'll follow up on that.

Our biggest fear going to the NIH was that we'd discover some not so good news, especially about the kidneys. Or, that we would be stuck in an unpleasant hospital stay which, as you know, carries significant emotional baggage for us. As it turned out, the kidneys looked fine despite his very high levels of MMA. The place was very different to a hospital stay, more like a mini vacation. Johnny did not want to leave. The NIH visit gave us a wonderful in depth and coherent picture of Johnny. This type of assessment of your child doesn't happen in the traditional doctor visit format. Really, no where else will you get such a meticulous discussion about the "what" but also "why". In all, we spoke for many hours about Johnny's results and about the research.

It is abundantly clear that Dr. Venditti shares a very special bond with these kids. We enjoyed our last with Dr. Venditti as we posed for pictures and spent time chatting. Johnny is really strong, a good wrestler which tickled Dr. Venditti since he was a champion wrestling in high school. It goes without saying that he is very special to us. He has committed his career to helping us find hope. He is truly our hero in the toughest struggle of our lives.

On March 26, we are holding a beef-n-beer fundraiser for MMA Research. For the flyer, info etc. please visit www.johnnytate.com

John and Lourdes Tate

Local Fundraiser for the Dr. Tanaka IVA Research Fund by Karen Bennett

At the end of October, we held a fundraiser to benefit the Dr. Kay Tanaka Isovaleric Acidemia (IVA) Research Fund and the Organic Acidemia Association (OAA). In a fun three hours, we raised over $1800.00. So many of our friends and family helped and came to the fundraiser.

I was so amazed and at one point was overcome by the amount of people that helped us and CARE to help. Once it began, every time I looked down the street, someone else was trying to find parking and walking to our house. People stayed a long time (despite cold weather) and commented that it was such a nice day. Most could not stay away from the hot dog/food stand that Melanie's Pop-Pop was in charge of, or the baskets of goodies that we were being raffled off by her Mima. These two events were two of the biggest money makers along with the moon bounce and kid activities. Many people also gave nice donations!

Many businesses help to contribute to such a successful event: Southern Living, Pampered Chef, PartyLite, Silpada Jewelry, Confections, Interior House Painting, Avon, Cassidy Video, Fresh Hair Studio, MaxWell Pharmacy, Slack's Hoagie Shack. We provided loads of fun for all including: Moonbounce Fun, Face Painting , Tractor/ Wagon Rides, Hot Dogs and Soda, Philadelphia Soft Pretzels, Baked Goods, Crafts to be made by kids including a coloring contest, and for the men---a Horseshoe Competition and Pie Eating Contest! We had several baskets to be raffled off including an Eagles Basket, a Fresh Hair Studio Basket, a Clinique Products Basket, and a Men's Basket.

It is a good feeling because some of the money is going to help Dr. Regina Ensenauer come to the OAA/ FOD Conference in June in Texas. She will be speaking about the IVA research she is doing. We actually had Dr. Ensenauer do the study for us, so it is a double good feeling!!

Melanie, IVA, enjoys wagon rides with her friends during the fundraiser.

We will do this again next year, so stay tuned. To grant Bill's wishes though, we are going to have it in the spring this time.

Karen, Bill, and Melanie Bennett
Melanie, IVA, 5-1/2 years old
Southampton, PA
bennka@centennialsd.org

— FOR IMMEDIATE RELEASE —

MUHC and McGill Scientists Identify Gene for Debilitating Vitamin B12 Disease

Montreal, 30 November 2005—Scientists at the MUHC and McGill University have identified a gene responsible for a disease that impairs the body's ability to handle vitamin B12 and that may contribute to heart disease, stroke and dementia. The details of the CIHR and March of Dimes funded research are published in this week's issue of Nature Genetics. The research, which began more than 20 years ago, will allow doctors to perform earlier diagnosis, assess 'carriers' of the disease—Combined Methylmalonic aciduria (MMA) and Homocystinuria—and open the door to new and improved treatments for this debilitating disease.

"Although this disease sometimes starts in adolescence or adulthood, we usually diagnose this rare inability to process vitamin B12 in the first few months of life," says Dr. David Rosenblatt, Chairman of Human Genetics at McGill, Director of Medical Genetics in Medicine at the MUHC, Chief of Medical Genetics at the Jewish General Hospital and lead researcher of the new study. "Babies may have breathing, feeding, visual and developmental difficulties, older patients may develop sudden neurological disease."

Vitamin B12, which is found in all animal products—including dairy, eggs, meat, poultry, and fish—but not in plants, is vital for synthesis of red blood cells and maintenance of the nervous system. Vitamin B12 also helps control homocysteine levels in the human body. Homocysteine control is important because in excess this compound can increase the risk of heart disease, stroke, and dementia.

17 year-old Michael—a typical MMA and Homocystinuria patient—was diagnosed at 6-months of age, and has battled numerous medical challenges as a result of his condition. Michael is developmentally delayed, visually impaired and does not talk; he has suffered seizures since he was three years old, had a stroke by the age of seven and has since developed rheumatoid arthritis and scoliosis. Michael's diagnosis, which led the way to treatment involving injections of vitamin B12, was conducted at Dr. Rosenblatt's laboratory at the MUHC—one of only two centres in the world that perform these tests.

After more than 20-years of data collection, Dr. Rosenblatt, his student Jordan Lerner-Ellis and their team have now unlocked some of the secrets of this rare but debilitating condition. "Using over 200 patient samples, representing the majority of the world's 350 known cases, we have identified the responsible gene, called MMACHC," says Dr. Rosenblatt. "In collaboration with the laboratory of Dr. James Coulton, Department of Microbiology and Immunology at McGill, we used computer modeling to demonstrate the similarity between the protein encoded by the MMACHC gene and a protein involved in bacterial vitamin B12 metabolism." This new link between bacterial and mammalian species may help us better understand how humans use vitamin B12. Ultimately these discoveries have enabled us to develop early diagnosis and carrier assessment tests for the disease—something that was not previously possible.

"This discovery offers earlier diagnosis and treatment options for genetic diseases such as Methylmalonic aciduria and Homocystinuria. This represents a step toward improving the lives of those afflicted with such rare and devastating genetic diseases," says Dr. Roderick McInnes, Scientific Director of CIHR's Institute of Genetics. This breakthrough represents hope for Michael and his family, and many others that have been touched by this disease. "Michael is a very loving and caring child, who has had to overcome many challenges," says his mother Karen. "We are overjoyed that this research may one day give courageous children like Michael a fighting chance at a better quality of life."

About medical genetics:

Alterations in our genes are responsible for thousands of hereditary diseases and influence the development of thousands more. Once the genes involved in a particular disease are discovered, scientists become better able to precisely diagnose disease, predict its course, and create more effective treatments with fewer side effects. Medical genetics can even be used to assess patients' risk of developing certain diseases, allowing them to take preventive medicines and make lifestyle changes, like diet and environment, which may help prevent or delay their development.

Medical genetics research is advancing at an incredible rate. This year alone, MUHC scientists have identified genes contributing to breast cancer, colon cancer, lung cancer, tuberculosis, migraines, cytomegalovirus (associated with herpes, chicken pox and mononucleosis) and rare but devastating diseases such as retinitis pigmentosa. The current work on Methylmalonic aciduria and Homocystinuria was a product of the CIHR group in Medical Genetics, comprised of scientists at the MUHC, McGill University, the University of Calgary and collaborators at the Hospital for Sick Children in Toronto.

The McGill University Health Centre (MUHC) is a comprehensive academic health institution with an international reputation for excellence in clinical programs, research and teaching. The MUHC is a merger of five teaching hospitals affiliated with the Faculty of Medicine at McGill University—the Montreal Children's, Montreal General, Royal Victoria, and Montreal Neurological Hospitals, as well as the Montreal Chest Institute. Building on the tradition of medical leadership of the founding hospitals, the goal of the MUHC is to provide patient care based on the most advanced knowledge in the health care field, and to contribute to the development of new knowledge. www.muhc.ca

McGill University is Canada's leading research-intensive university and has earned an international reputation for scholarly achievement and scientific discovery. Founded in 1821, McGill has 21 faculties and professional schools, which offer more than

continued on page 9, MUHC and McGill

MUHC and McGill, continued from page 8

300 programs from the undergraduate to the doctoral level. McGill attracts renowned professors and researchers from around the world and top students from more than 150 countries, creating one of the most dynamic and diverse education environments in North America. There are approximately 23,000 undergraduate students and 7,000 graduate students. It is one of two Canadian members of the American Association of Universities. McGill's two campuses are located in Montreal, Canada. www.mcgill.ca

The March of Dimes is a national voluntary health agency whose mission is to improve the health of babies by preventing birth defects, premature birth and infant mortality. Founded in 1938, the March of Dimes funds programs of research, community services, education, and advocacy to save babies and in 2003 launched a campaign to reduce the rate of premature birth. For more information, visit the March of Dimes Web site at www.marchofdimes.com.

The Canadian Institutes of Health Research (CIHR) is the Government of Canada's agency for health research. CIHR's mission is to create new scientific knowledge and to catalyze its translation into improved health, more effective health services and products, and a strengthened Canadian health care system. Composed of 13 Institutes, CIHR provides leadership and support to close to 10,000 health researchers and trainees across Canada. www.cihr-irsc.gc.ca

For more information please contact:

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Marie-France Poirier
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(613) 941-4563

Michele Kling
Director of Media Relations
March of Dimes
(914) 997-4613

For further information about Methylmalonic aciduria (MMA) and Homocystinuria, and for patient support, please contact:

Organic Acidemia Association (OAA):
Tel: 763-559-1797
Fax: 763-694-0017
www.oaanews.org

Coriell Institute for Medical Research

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.

The collection of samples from individuals with organic acidemias and other inborn errors of metabolism by the Human Genetic Cell Repository (HGCR) is for the establishment of cell lines. Cell lines contain live cells that can be used to study cellular functions. DNA can also be produced from cell lines. The most important thing about a cell line is that it is a renewable source of material, meaning that additional cells and DNA can continuously be made. Cell lines established at HGCR 30 years ago are still being used for research conducted today. HGCR's commitment to strict quality control practices will ensure that cell lines established today from individuals with inborn errors of metabolism will be available for researchers in future generations. These samples may even be used in the future as quality control material for DNA testing to ensure that laboratories across the country are performing quality diagnostic testing for inborn errors of metabolism. Lastly, the availability of these samples from a public repository will allow all researchers to have access to these materials, including ones that may not have studied these disorders otherwise due to the time and expense needed to collect their own samples.

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.

Editor's Note - Melissa's blood samples were sent last month to this important research.

Carbaglu Study for MMA/PA

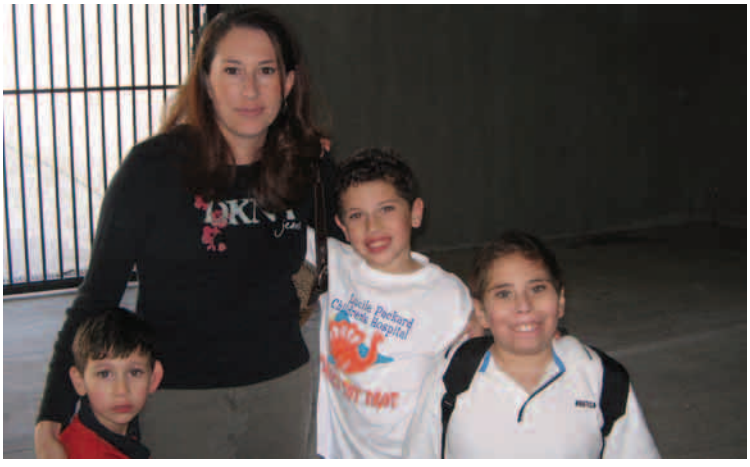
Children's Medical Center in Washington DC is recruiting MMA/PA patients to participate in a study on the drug "Carbaglu". The study is being conducted by OAA Medical Advisor, Dr. Mendel Tuchman. You may recall that he wrote an article on this study/drug in last summer's OAA newsletter. He is looking for a few MMA's and PA's who have elevated ammonia levels frequently higher than 50. These patients in all probability would have presented in the first few days of life with hyperammonemia. The drug is suppose to be helpful in keeping ammonia levels down in MMA and PA. This is an experimental drug and it has helped patients with urea cycle disorders. I have documentation on the study and contact information. Please email me privately if you are interested in learning more at oaanews@aol.com.

Update: Vincent Sanchez, MMA, Mut 0, Age 8
(Post Liver and Kidney Transplant)

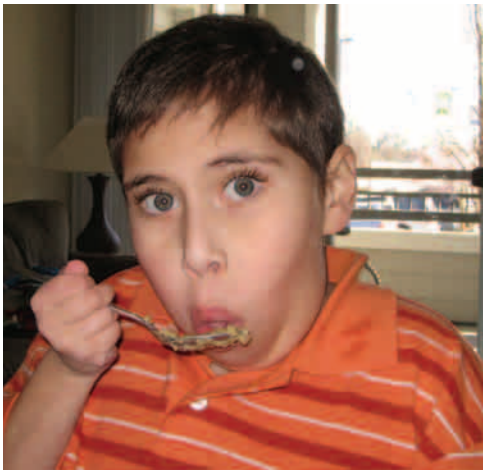
Vince's story began on February 19, 1997; some of you may have read the article on him about seven years ago. For those of you who didn't, I'll go ahead and reintroduce him. He was born in respiratory distress and the doctors caught onto the fact that he had MMA mut-0 way before he got really sick. They immediately started to treat him and avoided any major episodes. I'd like to say we lived happy and healthily ever after but unfortunately we didn't. Vince was sicker than most kids with MMA, he's never been able to go to school, caught a slight bug and ended up in the hospital, got excited and he would get sick. The truth is that he has struggled with his health since day one. Despite all the hard times he has added so much joy and love into our lives and has brought a new perspective and understanding of people in general. He has taught us to let life live and to set our lives by the old adage "don't sweat the small stuff".

Throughout his 9 years his MMA levels have been extremely high and we have tried everything to lower them to no avail. His kidneys started on a downward slope over the past 6 years and just continued that way until this summer when they stopped functioning properly all together. In July, we thought he had the flu when we found out that indeed it was his kidneys failing. Our docs called and told us we needed to start dialysis and had to get a hemodialysis catheter placed immediately. We went in on a Monday and had dialysis the following day. During this earth shattering experience and loss of hope we met with our metabolic doctor Johan Van Hove who told us that we could start to hope again. He explained that there are a few doctors who are treating MMA kids by removing the entire liver and the possibilities that can arise from the procedure. We felt that hope was returning and that we had no choice but to try it. The problem was that neither they nor we felt comfortable doing it back home in Denver. We had to go to the center where they had transplanted combined kidney and liver in two other patients with MMA, Stanford Medical Center. So we met with the docs at Stanford and decided to go ahead with the procedure. We were devastated to find out that he needed a cadaver donor and could not use one of us; we were overwhelmed with the fact that someone would need to die to save him. I don't think that we will ever get over the thoughts that go through our heads when we really stop to think about it. I can only say that we never wanted anyone to die and have only prayed for the best for the family of the donor.

We went on the waiting list November 29th, the waiting was painful but preparations were in order so we went into a robotic mode. On December 29th we went to a higher status and became number one on the waiting list. We prepared with work, school and insurance issues. On January 15th 2006, we received the call at 5:55 am. We immediately knew who it was. We hopped the next flight which was delayed by half an hour just for us. We arrived at Palo Alto at 10:30 and started the procedure. The fear was overwhelming and at times I just wanted to shout, "stop!" We started a 4-hour dialysis treatment and waited for the organs to arrive. Finally after a long sleepless night of waiting Vince went into surgery at 6:00 am Sunday January 15th, a day I will describe as one of the saddest yet happiest days of my life. The day seemed endless, we knew that 12 hours of surgery would but the wait was almost unbearable. Finally the first doctor came out and the liver was placed, he explained that it was a perfect fit, which is a good thing since they didn't have to cut down the liver at all. However, the spleen ruptured at the time they clamped his old liver and they tried to save it but could not. Another couple of hours and finally Dr. Conception and Dr. Benoit came to us and told us it was over, things went well. The kidney was placed and functioning. Thank God!



Vincent's Mom Lori, with brothers, Mikey and Evan.



First time Vincent eats in seven years (after transplant.)

The surgery was a huge success and Vince was recovering at record speed. The nurses and the doctors all said that they couldn't believe how fast things were moving along. After a week he was so well that they moved him up to the third floor to recover. Things were going great and then the crushing pain began. Vince was crying out in excruciating pain for two days before the doctors discovered that his hematocrit was dropping quickly and it must be because he is bleeding internally. An MRI showed that he was bleeding out from somewhere in the abdominally area and it needed to be stopped immediately. Emergency surgery was necessary at that time and the fear started all over again. Later to find out that this is fairly common after liver transplants. Vince's second surgery was only about an hour long and was very successful. He began recovering again and was transferred back up to the third floor after three days.

One afternoon were enjoying Vince and how well he was recovering while the doctors were telling us only a couple more days before we would go home, we had yet another scare. Vince had a grand mal seizure out of the blue, he stopped and then he started another one. He was immediately sedated and sent to have a cat scan to check for a stroke or any other abnormality of the brain. During the CAT scan he began another seizure and was sedated even more. They transferred him back to the room to review the results from the CT, he began another seizure and this time decided to stop

continued on page 13, Vincent Sanchez

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

http://www.oaanews.org
Organic Acidemia
Association

In order for us to serve you better, please fill out form COMPLETELY and Mail to Deb with your check (made out to OAA) By May 1, 2006

2006 FOD and OAA METABOLIC CONFERENCE REGISTRATION FORM

Location: Adam's Mark Hotel 400 North Olive Street, Dallas, TX 75201 for FOD & OAA sessions
http://www.adamsmark.com/dallas/index.asp
Call for Hotel Reservations: (214) 922-8000 (each Family/Professional makes their own reservation)

****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/Prof 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)

Registration Fee:

Number of people attending_____ @ $50.00 per person ($30.00 if you have an FOD/OA)
$_____enclosed (checks made out to OAA).
There will be **no daycare provided** for this Conference. Mature Teens and Adults are encouraged to attend.

_____Total Amount Enclosed (checks made out to OAA – mail to Deb (see next page)

_____I cannot attend, but please accept this donation for the meeting.

_____Volunteer to help with Conference ___Yes ___No

Families and Professionals: **___ # attending Thursday night Reception**
___ # attending Friday FOD or OAA session (circle one)
___ # attending Saturday half-day Joint session
___ # attending Saturday Tour of Institute (see below)

• • • Registration Fees cover the cost of meals and a special FOD or OAA T-shirt - Continental Breakfast will be served on both Friday and Saturday if you are staying at Adam's Mark. Lunch will be served on Friday only for all conference registrants. If you're not registered for the conference, then you won't be served a meal.

Name of Registrants (indicate which organization you are a member of – and state disorder)

Name:_____ **FOD/OAA?:_____**	**Tshirt Size?**	**Name:_____** **FOD/OAA?:_____**	**Tshirt Size?**
Name:_____ **FOD/OAA?:_____**	**Tshirt Size?**	**Name:_____** **FOD/OAA?:_____**	**Tshirt Size?**
Name:_____ **FOD/OAA?:_____**	**Tshirt Size?**	**Name:_____** **FOD/OAA?:_____**	**Tshirt Size?**

If you would like to order extra shirts for other familymembers, they are $10.00 per shirt **(separate check made out to OAA and mailed to Deb, along with this form by May 1, 2006)**

 # of__ small __ med __ lg __ xlg __ xxlg ____ FOD shirt or ___OAA shirt

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

Name_____**Address**_____

City_____ **State**_____ **Zip**_____ **Phone # ( )**_____

Email: _____

• • •

PROFESSIONALS: Please provide us with the following information:

Name _____ **Title** _____

Institution _____ **City, State** _____

Phone # _____ **Email** _____

Tentative Agenda

• 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)

For Agenda and Speaker Updates refer to: www.fodsupport.org or www.oaanews.org

TOUR of the Institute of Metabolic Disease (Saturday 2 – 4pm): A shuttle van will be provided to/from the hotel to take us to tour the Institute where Expanded Newborn Screening and diagnostic testing is performed. Dr Charles Roe, Medical Director, and Dr Larry Sweetman, Lab Director, will host.

• • •

SEE YOU THERE!

Kathy Stagni, Director Organic Acidemia Association
13210 35th Avenue North
Plymouth, MN 55441
(763) 559-1797 oaanews@aol.com

Deb Lee Gould, MEd, Director
FOD Family Support Group
1559 New Garden Rd, 2E
Greensboro, NC 27410
(336) 547-8682 deb@fodsupport.org

••• Mail Registration Form and OAA Check(s) to Deb's address •••
(I have a locked mailbox)

Organic Acidemia Association Friday Schedule

8:30-8:45 Introductions: OAA (Room TBA)

8:45-9:45 William Rhead, MD, PhD, Children's Hospital of Wisconsin
TOPIC: 'Late complications of OAs, liver transplants, and what the future will be'

9:45-10:00 **BREAK**

10:00-11:00 Charles Roe, MD, Institute of Metabolic Disease, Dallas, TX
TOPIC: 'New Concepts & Alternative Therapies In Inborn Errors of Metabolism'

11:00-12:00 James Gibson, MD, PhD, University of Texas Health Science Center, San Antonio
TOPIC: 'Developmental Issues with OAs'

11:45-1:00 LUNCH (Provided) together with FOD in Ballroom TBA

1:00-2:00 Barb Marriage, PhD, RD, Ross Products Division
TOPIC: 'How do I keep "Joey" following this DARN diet... and other burning questions about Organic Acidemias'

2:00-3:00 Chuck Venditti, MD, PhD, Genetic Disease Research Branch, National Human Genome Research Institute, National Institutes of Health –
TOPIC: TBA

3:00-3:15 **BREAK**

3:15-4:00 Jennifer Sloan PhD, MS, CGC, Protocol Coordinator & Genetic Counselor, NHGRI/NIH, **TOPIC:** 'Sibling Issues'

4:00 - **5:00** Regina Ensenauer, MD, Pediatric Research Center, Dr. von Hauner Children's Hospital, Ludwig-Maximilians-University Munich, Germany TOPIC: 'Isovaleric Acidemia: New aspects of genetic and clinical variability'

Saturday Schedule: Combined Session

8:15-8:30 Introductions (Room TBA)

8:30-9:30 Speaker: Gary Siskin, MD, Radiologist, Albany (NY) Medical Center and SCAD Dad, **TOPIC:** 'Tubes and Lines' [indications for use of g/gj tubes, PICC lines, ports, Hickman catheters etc; techniques of placement, potential issues with the tubes/lines, skin care etc; and solutions for these issues]

9:30-10:30 Speaker: Charles Roe, MD, **TOPIC:** 'Carnitine Issues and Metabolic Disorders'

10:30-10:45 **BREAK**

10:45-11:45 Speaker: Brad Therrell, PhD, National Newborn Screening and Genetics Resource Center, **TOPIC:** 'The World of Newborn Screening'

11:45–1:45 Sideshow/Ribbon Ceremony and then LUNCH on own, by 1:45meet outside hotel for bus ride to Institute 2-4pm Tour of the Institute of Metabolic Disease - Buses will drive us to/from the Institute –10 min drive

Speaker Times are Subject to Change

Vincent Sanchez, continued from page 10

breathing. They rushed him to the ICU and immediately began working on him. This day is the most devastating time of my life and I still have nightmares from it. They yelled at him to respond to them and he wouldn't. We had three ICU doctors, the head of the ICU, two surgeons and many nurses working on him. As I looked into the room at him lying there lifeless, I asked if he was dead, no one answered me yet. You know things are bad when the chaplain is paged and he is sitting next to you. We prayed and prayed for things to be ok but still weren't sure if they were. Finally, doctors started to sigh a little and started to leave, they had saved him. However, we still didn't know to what degree. The neurologists were called and couldn't give us answers; they just had no idea if he would wake up or not. His fragile life laid in the hands of God now we had no control anymore. They hooked him up to the EEG machines and let him be, he would not respond to anything including pain. Aaron and I were completely exhausted and overwhelmed so we took shifts to sleep. At 2:30 am Aaron woke me up and told me that my little boy was asking for his mommy. It was truly the happiest day of my life he was back. Once again, Thank God! He made a full recovery although the doctors still admit that they were pretty scared! We found out that the seizures were probably from a high level of immune suppressant drug called Prograf. His level had been high the day before but we still don't know for certain so we still have him on Keppra for the seizures. One good thing we found out is that his brain is that of a normal 9-year-old boy, something I can't say we expected.

So now, we are still very much in the recovery process. Everyday seems to get a bit easier as we face new challenges. Vince is definitely happier and we have noticed major changes in him. He seems so alive. Our prayers continue to go out to the donor family and we thank them in our hearts every time we look at Vince.

One final note to give the logistics. Pre transplant MMA 3500, post 74. Pre creatinine 5.2, post .63. Lactic acid, pre 5.2, post 1.2. Protein intake pre transplant .75 (and that was pushing it) post 1.4. Things are definitely better.

God bless you all !

Lori Sanchez and family
6260 W. Sumac Avenue
Littleton, CO 80123
303-798-5023
loriasanchez@msn.com

Apples to Zucchini: A Collection of Favorite Low Protein Recipes

by Virginia Schuett and Dorothy Corry

Here is another sample page from our new cookbook, Apples to Zucchini. We hope it will inspire you! These are just two of the 562 exciting recipes you will find in a book that emphasizes naturally low protein foods rather than special low protein products.

Taken from the National PKUNews newsletter, Vol. 17, Number 3 Winter 2006.

Quick Curried Cauliflower and Potato Soup

The curry in this warming, Indian-inspired soup is mild; add more to taste. You can make it into a fully puréed soup, or if you prefer, you can just purée part of the soup and leave some chunks of cauliflower and potatoes (my preference). An immersion blender works well for the chunky version. VS

- 3 tablespoons canola oil
- ½ medium onion, chopped (120 gm)
- 1 tablespoon sugar
- 1 tablespoon curry powder
- 1 medium head cauliflower, cored and cut into small florets (460 gm)
- 2 medium Yukon Gold or white potatoes, peeled and cut into ½-inch dice (270 gm)
- 5½ cups vegetable broth, canned or homemade
- salt and pepper
- chopped fresh flat-leaf parsley for garnish (optional)

1. Heat oil in a large saucepan or stockpot over medium-high heat. Add onion and sauté until tender, about 4 minutes. Add sugar and curry powder; stir for 1 minute. Add cauliflower and potatoes; stir for 1 minute.
2. Add broth and bring to a boil. Reduce heat to low, cover, and simmer until vegetables are tender, about 30 minutes.
3. Purée soup in a food processor or blender in batches as needed. (Do not fill blender more than half full.)
4. Return soup to the pot and bring to a simmer. Season to taste with salt and pepper. Ladle soup into bowls. Sprinkle with a little chopped parsley if desired.

Quick Curried Cauliflower and Potato Soup

Makes 7 cups
½ cup per serving

	recipe	serving
protein (gm)	16.4	1.2
calories (kcal)	977	70
phenylalanine (mg)	606	43
tyrosine (mg)	403	29
leucine (mg)	892	64
sodium (mg)	3076	220
potassium (mg)	2934	210
phosphorous (mg)	410	29

Southwestern Pumpkin Soup

Almost as easy to make as opening a can of soup, this unusual and delicious soup is the perfect starter for a fall meal. Serve topped with Chili Croutons, page 119, for a crunchy kick. For non-diet portions, you can top the soup with grated cheddar cheese or a dollop of sour cream. To change the flavor completely, try making it into Cinnamon-Spiced Pumpkin Soup by replacing the cumin, coriander, and chili powder with ½ teaspoon each ground cinnamon and ground nutmeg; serve this version topped with Cinnamon Croutons, page 119. VS

- 2 cups vegetable broth, canned or homemade
- 1 cup water
- 1 cup liquid non-dairy creamer
- 1 15-ounce can pumpkin (425 gm)
- 2 tablespoons packed brown sugar
- 1 teaspoon ground cumin
- 1 teaspoon ground coriander
- ½ to 1 teaspoon chili powder, or to taste
- salt and pepper
- chopped fresh cilantro for garnish (optional)
- Chili Croutons, page 119 (optional)

1. In a medium saucepan or stockpot, bring broth, water, and non-dairy creamer to a boil over medium-high heat. Mix in pumpkin, sugar, and spices.
2. Reduce heat to medium-low and simmer, uncovered, until soup thickens a little and flavors have blended, about 10 minutes. Season to taste with salt and pepper. Serve garnished with chopped cilantro and Chili Croutons if desired.

Southwestern Pumpkin Soup

Makes 5 cups
(not including croutons)
½ cup per serving

	recipe	serving
protein (gm)	5.5	0.5
calories (kcal)	628	63
phenylalanine (mg)	181	18
tyrosine (mg)	228	23
leucine (mg)	277	28
sodium (mg)	2115	211
potassium (mg)	1021	102
phosphorous (mg)	155	15

To Purchase the Book

You can conveniently place an on-line order with SHS North America by going to myspecialdiet.com, then clicking on Product Information and On-Line Shop. Or use the order form on the purple page (page iv) or call SHS at 1-877-482-7845. US residents: $37 US plus $10 shipping for a single book ($6 shipping for 2-4 books, $5 shipping for 5 or more books). Non-US residents: Please call SHS at 1-877- 482-7845 for shipping cost.

What People Are Saying about Apples to Zucchini

• Apples to Zucchini is a masterpiece. . .a wealth of information on low protein cooking that is sure to become a "bible" for those who follow low protein diets. With a lovely, soothing, and easy-to-read layout, it is a combination of both art and science. . .the art of creative cookery coupled with the science of low protein nutrition and healthful eating which focuses on incorporating wholesome foods into medically necessary diets.
—Fran Rohr, MS, RD, Childrens' Hospital, Boston, MA

• Apples to Zucchini is the only cookbook that has gotten me consistently excited about making dinner every night. As a physician, a vegetarian, a mother of three, and most importantly, the mother of a child with PKU, this book is a dream come true.
—Emily Brandenfels, MD, Seattle, WA

• Apples to Zucchini is a wonderful cookbook. My son has always been a picky eater, but since getting the book recently, he has already tried several recipes that he loves. The book has tons of post-it notes throughout, highlighting recipes that he wants to try. I finally feel I can feed and satisfy him with things other than fries, fried dough, and pasta!
—Nancy Santerre, Thompson, CT

• I have lived with PKU for 33 years and this cookbook is a welcome new development. It opens the door to a variety of new and exciting dishes for the whole family, not just for those whose diets are restricted. The heart of the cookbook is the sheer number and variety of recipes, which conquers the myth that low protein diets are boring.
— John Reyzer, Waunakee, WI

• I just wanted to tell everyone how incredibly amazing Apples to Zucchini is! It is a beautiful cookbook that gives new life and inspiration to the challenge of living with the PKU diet. I especially appreciate their use of whole, natural foods rather than expensive and highly processed specialty foods. I love that the recipes can be made for the whole family.
—Sally Haugen, Boulder, CO

• This book is a gift to those of us who work with persons who require a daily protein restriction to remain healthy. The most wonderful aspect of the book is that all of the recipes use "real food"; that is, they use a wide variety of fruits and vegetables that support good health and nourishment for all of us. The recipes are exciting to read and even more fun to actually cook.
—Cris Trahms, MS, RD, PKU/ Biochemical Genetics Program, Seattle, WA

Cambrooke Foods Update

Try our new products! Scoop and bake Sugar Cookie Dough - use to roll, decorate and bake or just scoop and bake right out of the container. We had many requests for plain Cheese Ravioli and our tasting panel is thrilled with the results. Joining our line of fresh filled frozen pastas is Cheese-filled Ravioli, now available to order. All of our delectable filled pastas are versatile products that can be used "as is" or with your favorite sauce. The new Cambrooke Foods' White Cheddar Orzo is a no-artificial-coloring alternative for Macaroni and Cheese lovers.

Understand better how your diet and protein supplements are affecting your wellness with DietWell™, Cambrooke's easy to use Dietary Wellness Network. Record and monitor daily protein and supplemental protein intake, see the relationship between your diet and your wellness. Receive personalized reports tracking your diet and wellness - perfect for bringing to clinic visits. Patients can receive Cambrooke Foods' "DietWell™ Points", good for the purchase of your favorite Cambrooke Foods' products as an incentive to monitor your diet better.

Do you like to receive weekly recipes and special promotions? Make sure Cambrooke has your correct e-mail address and we will send these out to you. See our posted recipes on our website under the "Recipes and Tips" tab.

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

Update from Applied Nutrition

by Sarah Foster, adult PKU

Over the last several years, great strides have been made in both the quality and variety of low protein foods. I can remember as a kid growing up with PKU and having only two modified low protein food choices available; Aproten Spaghetti and wheat starch. My mom would work so hard to make low protein pizza, but it always looked and tasted, like a brick to me. Thankfully, those days are long behind us!

Even with the advances and improvements to low protein foods in recent years, there still was no low protein food that satisfied my desire for a salty/savory snack. I had been looking for something to suit my craving that didn't "break the bank" when it came to my protein allotment for the day. I wanted a low protein snack that was salty and good. I am a lucky person because I had the ear of someone who could actually do something about this "problem".

As many of you know, I am a sales representative for Applied Nutrition Corporation. The person's ear I had was Rick Finkel, the owner and president of Applied Nutrition. Rick is a great person and always willing to help when he is able.

After listening to me express my views that the metabolic community would like a salty/savory snack food, Rick and Sandra Maltzman, the registered dietitian for Applied Nutrition Corp., went to work in the lab developing the first ever commercially available savory snack food specially formulated to have less than 1 gram of protein per serving.

I'm happy to be able to let the OAA community know that Rick and Sandy's work has resulted in a new low protein product called Tangles®, by Maddy's®. Tangles® are the little puff with the big crunch. They have the crispy texture and zesty taste we've been looking for. Available in three delicious flavors; Smokin' Bar-B-Q, Salt & Vinegar and Original. They are packaged in a convenient 1? ounce Jumbo Grab bag with only 0.4g of protein and 0.25 mg of Leucine per bag. They look just like a snack grabbed off the shelf at your local convenience store but fit the low protein diet!

If you'd like more information about Tangles® simply visit www.DietForLife.com or call 1-800-605-0410 and Nichole or Sonja will be happy to help you.

Easter Treats from Applied Nutrition

Cedar Knolls, NJ - March 2006

Make your Easter a little sweeter this year with Maddy's® Energy Option® treats for your holiday basket. Available now through Easter, Applied Nutrition is proud to offer protein free confections in Chocolate and White Chocolate flavored holiday shapes. Protein free solid bunnies, protein free bunny pops in three color varieties, and protein free bite size holiday colored Easter treats.

Supplies are limited so place your order soon for this holiday season. Applied Nutrition is offering complimentary shipping on these and all other Maddy's® products, including the recently introduced low protein snack chip, Tangles®.

As a recognized leader in metabolic nutrition, Applied Nutrition Corp. has always been committed to providing great tasting, easy to use medical food products. The company sells product nationwide and in 13 countries around the globe with existing brands PhenylAde® and Complex MSUD®. By incorporating existing Energy Option® products into the Maddy's® brand consumers continue to have more variety in choosing products they can truly enjoy.

To learn more or to print an order form for Maddy's® Easter treats and other low protein products, please visit us on the web at www.dietforlife.com. For more information and free samples of our medical food products, please call 1-800-605-0410 or visit us at www.medicalfood.com.

Applied Nutrition is dedicated to the needs of the metabolic community and those that serve them, if you have any events scheduled and would like product samples & literature don't hesitate to contact us at 1-800-605-0410 or info@medicalfood.com.

Please forward this message on to anyone you think may be interested.

Sincerely,
Sonja O'Brien

Check out OAA's website for ordering forms: http://www.oaanews.org/EasterTreats.htm

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