Organic Acidemia Association
www.oaanews.org

Newsletter

3175 Ironhorse Dr, Woodbridge, VA 22192
Voice: 703-497-1216 • Fax: 866-539-4060
jana.monaco@verizon.net

Winter 2009 Volume XXVI, Issue 1

WHAT'S INSIDE

Happy New Year

Happy New Year! This edition of the OAA Newsletter is the first for 2009 and the first for me as the new Executive Director of OAA. My name is Jana Monaco and I am the mother of four children to include Nicholas, 17 ½, Alex, 15, Stephen 11 and Caroline 6 years old.

It seems like just yesterday, when my husband Tom called Kathy to introduce himself and inform her that our son Stephen, then 3 ½, experienced a metabolic acidosis and was given a diagnosis of Isovaleric Acidemia. It was no doubt a very difficult and trying time for us as a family as we struggled to accept the disorder and the neurological damage as a result of a late diagnosis. Connecting with other families in OAA has been incredibly comforting and empowering to us. Receiving a diagnosis of one of these disorders can be tremendously lonely, but the loneliness quickly left us as we bonded with the OAA families. It did not take long before OAA became our extended family and helped us welcome our daughter Caroline just a year later, who was also born with IVA. An early diagnosis, prior to birth, protected her from the acidosis that Stephen experienced.

The past seven and a half years have taught me not only about organic acidemias, but caring for a child with complex health problems and disabilities. I have advocated for newborn screening on the state and national level and serve as a member of the HRSA Advisory Committee for Genetic Diseases in Newborns and Children. My personal experiences and knowledge will no doubt serve as a firm foundation for me in my new role as executive director.

My goal is to continue the amazing work that Kathy Stagni has done for OAA over the many years that she has served as executive director. We have all benefited from the camaraderie and education that is found with OAA and many can attest that such support was not available to families years ago. With the advancements made with these disorders, so has the network of people to turn to along with the vast array of information. We are scattered around the globe, but are fortunate to connect through OAA.

To Kathy, I extend a sincere thank you for all of your hard work and dedication to families with organic acidemias and am glad that you will remain working with our board in a different and less demanding role.

I am please to announce our Board of Directors that will assist me in fulfilling the mission of OAA:

Executive Director-Jana Monaco, mother of Stephen and Caroline (IVA)

Finance Director-Marty Moran, father of Kayleigh (MMA Mut 0)

Research Director-Janice Boecker, mother of Kristin (PA)

Communications Director- Lori Sanchez, mother of Vince, (MMA Mut 0)

Administrative Director-Kathy Stagni, mother of Melissa (PA)

Family Support Director-Kerri Wagner, mother of Hudson and Hope, (IVA)

With the passing of another year, we celebrate each other's milestones, welcome new members and remember those who we have lost. I hope you will enjoy the informative articles contributed by several of our professionals in the field along with the personal articles about several of our kids, from families.

Jana Monaco

Board of Directors

The Organic Acidemia Association(OAA) provides information and support to parents and professionals dealing with a set of inborn errors of metabolism collectively called 'organic acidemias'. The OAA is a volunteer organization registered with the IRS as a 501(c)(3) non- profit corporation. Donations to the OAA are tax-deductible. OAA publishes a newsletter three times a year, hosts an internet-based list-serv for information exchange and maintains a website. These services are funded by donations from corporation and individual members. Annual membership donation of $25 (U.S) and $35 (international) plus $5 for the family roster is requested, but not required. Our 501(c)(3) non-profit status qualifies OAA for United Way donations through their write-in option. If there is a write-in option, just write "Organic Acidemia Association" in the blank line on your pledge card. Donations can also be made at OAA's website through the "PayPal" and the "Network for Good" option.

- 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.

MEDICAL ADVISORS

Dr. Olaf Bodamer FACMG
Biochemical Genetics and
National Screening Laboratories
University Children's Hospital
Vienna
Wahringer Gurtel 18-20
1090 Vienna, Austria

Stephen Cederbaum, MD
Mental Retardation Research
Ctr./NPI
635 Charles E. Young Dr.
South Rm 347
Los Angeles, CA90095-7332

Richard Hillman, MD
Univ of Missouri Health
Sciences Ctr
G-1 Metabolism Clinic
Department of Child Health
1 Hospital Drive
Columbia, MO 65212

Elaina Jurceki, MS, RD
Kaiser Permanente Medical Ctr
280 W. MacArthur Blvd.
Oakland, CA94611

Stephen G. Kahler, MD
Professor,
Division of Clinical Genetics
Department of Pediatrics
University of Arkansas
for Medical Sciences
Division of Clinical Genetics
Slot 512-22
Arkansas Children's Hospital
800 Marshall St.
Little Rock, AR 72202-3591

J.V. Leonard, MD
40ABagley Wood Road
Kennington Oxford OX1 5LY
United Kingdom

Dr. Seymour Packman, MD
Dept of Pediatrics, Box 0748
Univ of California,
San Francisco
San Francisco CA94143-0748

Piero Rinaldo, MD, PhD
Mayo Clinic
Lab Genetics
200 First St., S.W.
Rochester, MN 55905

Mendel Tuchman, MD
Vice Chairman for Research
Scientific Director, Children's
Research Institute, Professor of
Pediatrics, Biochemistry &
Molecular Biology
The George Washington
University School of Medicine
111 Michigan Avenue, NW
Washington, DC 20010-2970

Keiko Ueda, MPH, RD, LDN
Metabolic Dietitian
Department of Pediatrics
Division of Metabolism
Floating Hospital for Children
New England Medical Center
750 Washington Street,
NEMC#434
Boston, MA02111

Charles P. Venditti MD, PhD
Genetic Disease Research
Branch
National Human Genome
Research Institute
National Institutes of Health
Bldg 49, Room 4A56A
Bethesda, MD 20892-4472

Jerry Vockley, MD, PhD
Professor of Human Genetics
and Pediatrics
Chief of Medical Genetics
Children's Hospital of Pittsburgh
3705 Fifth Avenue
Pittsburgh, PA 15213
Dr. Susan Winter, MD
Valley Children's Hospital
9300 Valley Children's Place
C/O Medical
Genetics/Metabolism SE11
Madera, CA 93638

Arthur B. Zinn, MD, PhD
Center for Human Genetics
CWRU/University
Hospitals of Cleveland
11100 Euclid Avenue
Cleveland, Ohio 44106

PATIENT-AS-TEACHER PROJECT

At Floating Hospital for Children at Tufts Medical Center, Boston, MA

There is a shortage of metabolic physicians practicing in North America and not enough young doctors training in this subspecialty. Meanwhile, the field is advancing in many successful ways. Newborn screening for metabolic disease continues to expand, identifying more patients in need of attention and follow-up. New effective treatments are becoming available. Many patients are living longer and remain in better health; they need long-term metabolic care.

The Metabolism Service at Tufts Medical Center has launched the **Metabolism Outreach Service (MOS)**, the largest metabolic educational outreach program ever created, to several teaching hospitals in the northeastern U.S. It consists of lectures and workshops that highlight a metabolic approach to a wide range of symptoms, opportunities for patients and families to address medical audiences, and scheduled occasions to review patient cases with physicians when metabolic disease should be considered.

Metabolic education of physicians must be a priority so that they can participate more in the diagnosis and management of metabolic patients. Medical students need to hear about and see patients with metabolic disease. We need to get them interested so they might consider the area of genetic metabolic disease as a career choice.

An essential component of the Metabolic Outreach Service is the Patient-As-Teacher Project, and this is where you can play a part in one of two ways (or both!):

1. **Consider speaking to medical audiences** i.e., medical students at a medical school, or residents and attending physicians at a teaching hospital. Patients and parents can teach in a very compelling way that makes for a powerful message. Medical audiences also appreciate a patient's perspective on his/her journey through the health care system.

2. **Allow the Metabolic Outreach Service to review you/your child's medical record and use it as a teaching case.** Teaching is much more effective when it includes a real case; each case usually has many teaching points to offer. Having many cases available means the cases can be changed periodically in order to challenge those attending the metabolic workshops.

I***f you are interested in participating in the Patient-As-Teacher Project***, please review the attached forms at this website: http://www.floatinghospital.org/OurServices/Genetics_Metabolism/PatientAsTeacherProject/MOSBOTHNEWEST.doc and mail them in or fax them back to our office.

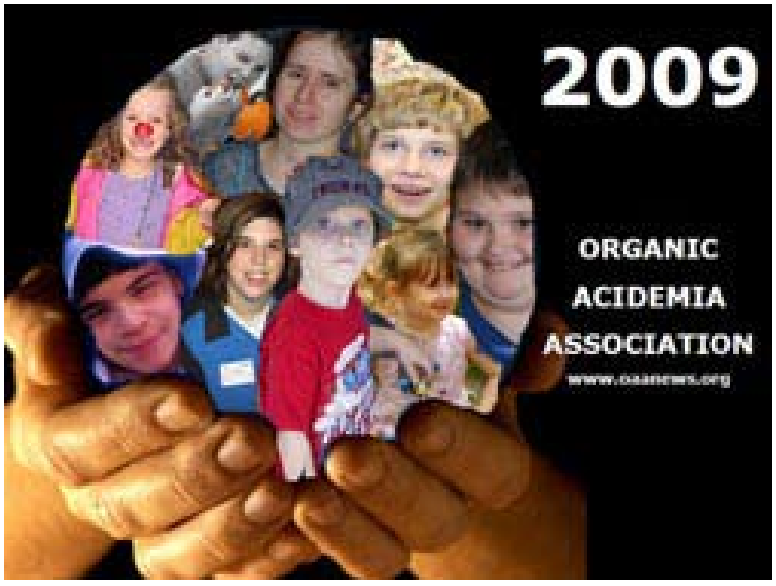
If you have any questions, please call (617) 636-5443.
There is so much work to be done in the area of genetic metabolic disease. Thanks for helping in this important effort.
Editor's note – Dr. Korson reports that he currently has 50 families representing 20 different disagnoses – please consider helping if you live in this area.

The METABOLIC OUTREACH SERVICE

The Metabolic Outreach Service (MOS) is an ongoing educational and consultative assistance program for academic medical centers that don't have an on-site metabolic service. The aim of the MOS is not just to increase awareness about metabolic disease but to change clinical practice, enabling clinicians to "think metabolic." Participating centers include Boston Medical Center (Boston, MA), Baystate Medical Center (Springfield, MA), Dartmouth-Hitchcock Medical Center (Lebanon, NH), Vermont Children's Hospital (Burlington, VT), Eastern Maine Medical Center (Bangor, ME), and Maimonides Medical Center (Brooklyn, NY).

After 3 years of planning, the Metabolic Outreach Service has finally begun! The MOS launch has been very successful, and thus has established a short term plan as well as a plan for the future. Our achievements thus far could not have been accomplished without the support of our sponsors, whose generosity we continue to rely on for the necessary funding to sustain and grow over the upcoming years.

Order your 2009 OAA Calender!



Available at CafePress.com:

http://www.cafepress.com/organicacidemia

Spotlight on Dietary Fiber & OAs

Keiko Ueda, MPH, RD, LDN, Metabolic Dietitian, Floating Hospital for Children at Tufts Medical Center & Emily Evans, MS, Frances Stern Nutrition Center Dietetic Intern, Tufts University Friedman Graduate School of Nutrition, Boston, MA

Fiber is primarily from plant sources and provides plants with their shape and structure. Dietary fiber refers to a group of complex carbohydrates that humans can not breakdown to use for calories as we lack the digestive enzymes. In the past because of increasing public preferences for softer white breads, grains were increasingly refined to make flours, but in the processing the flours lost fiber as well as other vitamins (iron, riboflavin, niacin and thiamin). Flours were then enriched to replace lost nutrients e.g. fiber, as well as fortified with added nutrients e.g. folate1. The 2008 American Dietetic Association (ADA) evidence analysis review of fiber studies2 indicate that there is some evidence that adequate dietary fiber intakes from foods or supplements may provide some protection from cardiovascular disease by lowering blood pressure, improving serum lipid levels, and reducing inflammation. But more studies are needed to clarify if fiber provides health benefits towards promoting weight loss; lowering blood sugar levels for people living with diabetes; and if fiber is helpful in colon cancer prevention; and prevention of gastrointestinal disorders such as chronic constipation, irritable bowel syndrome and diverticulitis.

Most Americans obtain their dietary fiber from white flour food products and potatoes, and the usual intake of fiber in the US is only 15 grams/day. The Food and Nutrition Board of the Institutes of Medicine recommends that an adequate fiber intake for adult women is 25 grams per day, for adult men is 38 grams per day or to aim for 14 grams of dietary fiber for every 1,000 calories consumed. Adequate fiber intake for infants aged 0 to 12 months old, the very old, and critically ill people is not determined3. The ADA's 2008 nutrition recommendations now include encouraging 'the public to consume adequate amounts of fiber from a variety of plant foods'2 such as whole grains, legumes (beans), fruits and vegetables. So how does this apply to people living with organic acidemias (OAs)?

High Fiber-Low Protein foods

Most people living with OAs are following a medically monitored and prescribed protein restricted diet. Some OA patients must only limit their daily amount of protein intake in foods; while others must also consume an individualized, OA disorder-specific metabolic formula (medical food). Table 1 lists selected foods and the fiber and protein content for a given serving size, data gathered from the USDA National Nutrient Database4. The table shows that most high fiber-rich foods such as whole grains and legumes are also higher in protein content, so that these high fiber foods are often avoided or limited in people following OA protein restricted diets. But many lower protein foods such as fruits and vegetables can also be a significant source of dietary fiber for people living with OAs. There is also a low protein food company currently enriching their low protein flour with added fiber, and many low protein bread recipes include the addition of psyllium fiber to improve texture of the low protein bread slices.

Table 1 Fiber and Protein in Selected Foods

Food	Serving	Fiber (grams)	Protein (grams)
Grains, Legumes, Nuts & Seeds			
All-bran, bran buds cereal (Kellogg's)	1/3 cup	12.9	2.3
Whole wheat spaghetti, cooked	1 cup	6.3	7.5
Brown rice, long grain cooked	1 cup	3.5	5
Raisin bran cereal (Kellogg's)	_ cup	3.3	2.5
Oat bran, cooked	_ cup	2.8	3.5
Spaghetti, cooked	1 cup	2.5	8.1
Whole wheat bread	1 slice	1.9	3.6
Popcorn, air popped	1 cup	1.2	1
White rice, long grain regular cooked	1 cup	0.6	4.3
White bread	1 slice	0.6	1.9
Navy beans, cooked	_ cup	9.6	7.5
Lentils, cooked	_ cup	7.8	8.9
Kidney beans, cooked	_ cup	5.7	7.7
Peas, green, cooked, boiled	_ cup	4.4	4.3
Almonds, dry roasted	1 ounce	3.3	6.3
Sunflower seed kernels, dry roasted	1 ounce	2.6	5.5
Fruits			
Persimmons, japanese raw	1 fruit	6	1
Avocado, raw, cubed	_ cup	5	1.5
Raspberries, raw	_ cup	4	0.7
Figs, dried uncooked	1/4 cup	3.7	1.2
Prunes, dried uncooked, pitted	1/4 cup	3.1	0.95
Pear, raw, slices	_ cup	2.2	0.3
Orange, raw sections	_ cup	2.2	0.85
Banana, raw sliced	_ cup	1.9	0.8
Blueberries, raw	_ cup	1.8	0.6
Strawberries, raw, sliced	_ cup	1.7	0.6
Apple, raw with skin slices	_ cup	1.3	0.1
Raisins, seedless	1 ounce	1	0.9
Vegetables			
Squash winter, all varieties baked	_ cup	2.9	0.9
Broccoli boiled, drained, chopped	_ cup	2.6	1.9
Spinach, boiled, drained	_ cup	2.2	2.7
Brussels sprouts, boiled, drained	_ cup	2	2
Potato, French fr, all types, frozen, oven heated	10 strips		
(74 grams)	1.9	1.97	
Carrots, raw, slices	_ cup	1.7	0.6
Cauliflower, boiled drained	_ cup	1.4	1.1
Squash, summer, all varieties boiled sliced	_ cup	1.3	0.8
Potato, baked with skin	_ cup	1.3	1.5
Tomato, red, ripe, raw, sliced	_ cup	1.1	0.8
Lettuce, romaine	1 cup	1	0.6
Lettuce, iceberg raw, shredded	1 cup	0.9	0.65
Cucumber with peel, raw, slices	_ cup	0.3	0.3
Celery, raw	4 inch strip (4 grams)	0.1	0.03

What is Fiber?

In the past, fibers were most often defined by their chemical properties; soluble fibers e.g. pectins, gums, mucilages that partially dissolve in water or insoluble fibers e.g. cellulose, hemicellulose, lignin that do not dissolve in water. Insoluble fibers may help with intestinal regularity, while soluble fibers may help prevent heart disease. Lower protein foods containing insoluble fibers include; raspberries, banana, apple, kiwi, tomato, cucumber, potato and lower protein foods with soluble fibers include apple, grapefruit, pears, broccoli, and potato1. Most foods contain both soluble and insoluble fibers. Therefore in 2002 the Institute of Medicine recommended we use the terms; dietary fiber, functional fiber, and total fiber to help future fiber research. Dietary fiber is the nondigestible carbohydrates and lignin that are intrinsic and intact in plants. Functional fibers are isolated nondisgestible carbohydrates that have beneficial physiological effects in human beings. Total fiber is the sum of dietary fiber and functional fiber3.

Regardless of fiber definitions, many people living with OA disorders often go through stages of eating limited amounts of foods by mouth, requiring additional metabolic formula tube feedings to meet their daily nutritional needs. People living with OAs who eat small amounts of food often have particular preferences for favorite foods not necessarily including fiber rich or nutritive food choices, just like most Americans. Formula manufacturers currently do not add fiber supplements to their OA disorder specific metabolic formulas. Therefore to provide a significant daily source of dietary fiber for people living with OAs who eat limited amounts of food by mouth, parents and clinicians may need to consider the addition of a fiber supplement.

Fiber supplements and OAs?

Formula manufacturers first started to add fiber supplements to their non-metabolic formulas assuming that it would help normalize bowel function and prevent constipation and diarrhea. The 2008 ADA evidence analysis of the health implications of dietary fiber2 concluded that there is a lack of studies to prove the health benefits of taking fiber supplements. There have also been a few case reports that excessive fiber intake may be harmful for some people resulting in excessive diarrhea or bowel obstructions. Excessive dietary fiber intake may also interfere with the absorption of some medications as well as nutrients such as iron, calcium and zinc. There is also the logistical possibility of some fiber supplements (e.g. gums) when added to formulas, thickening formulas then blocking enteral formula feeding tubes. Anne Pylkas and others in a 2005 fiber study5 compared various dietary fibers for short chain fatty acid (e.g. acetate, propionate, and butyrate) production. The researchers found that hydrolyzed guar gum and galactomannan produced the greatest amount of total short chain fatty acids; methylcellulose and arabinogalactan resulted in the most propionate production. It is important to note that this study was done in test tubes not with actual human participants living with OAs. But the results of this study suggest that there may be particular types of fiber supplements that are contraindicated for people living with propionic acidemia and short chain fatty acid metabolism disorders. Also if short chain fatty acid intestinal production is theorized to be helpful in intestinal function and health; the avoidance of these scfa promoting fibers may negate part of the health benefits of taking fiber in the first place.

Discuss Fiber with Your Clinic

There are more studies and public awareness about the probable health benefits of consuming an adequate amount of fiber in foods that may also apply to people living with OAs. But further studies are needed to prove the health benefits, clarify the amount of daily fiber and the type of fibers most appropriate for people living with OAs. It's known that anyone trying to increase dietary fiber intake should do so slowly over time with adequate fluids to lessen the acute symptoms of abdominal distention, bloating and flatulence. People living with OAs should always first consult their metabolic clinic physician and dietitian before adding fiber supplements and/or trying to increase dietary fiber intake in foods to best weigh the health risks vs. health benefits for their individual situations.

References

1. Roberta Duyff, MS, RD, FADA, CFCS. 'Fiber Your Body's Broom'. American Dietetic Association Complete Food and Nutrition Guide, 2nd Ed. John Wiley & Sons, Inc. Hoboken, NJ 2002

2. Joanne Slavin et al. 'Position of the American Dietetic Association: Health Implications of Dietary Fiber' Journal of the American Dietetic Association, Oct 2008;108:1716-1731

3. Institute of Medicine. Dietary Reference Intakes for Energy, Carbohydrate, Fiber, Fat, Fatty Acids, Cholesterol, Protein, and Amino Acids. 2005. Washington, D.C.: The National Academies Press. Website: http://books.nap.edu/openbook.php?isbn=0309085373

4. USDA National Nutrient Database for Standard Reference website: http://www.nal.usda.gov/fnic/foodcomp/search/

5. Anne Pylkas, Lekh Juneja, and Joanne Slavin. "Comparison of different fibers for in vitro production of short chain fatty acids by intestinal microflora" Journal of Medicinal Food, March 2005; 8(1):113-116

Connecting with OAA

OAA Internet Listserv

OAA's main mission is to empower families with knowledge about organic acidemias.....if you would like to connect with other families who share the same or similar diagnosis -- please join our OAA Listserv. Visit OAA's website to sign up at: http://www.oaanews.org/listserv.htm OAA currently hosts two Listserv - one for families and professionals and one for adults living with an organic acidemia.

OAA Inspire Community Forum

http://www.inspire.com/groups/organic-acidemia-association/
OAA joined with NORD to create our own "Inspire" Community Forum. Check out the link about to connect with OAA and a number of other rare disease groups.

OAA is on Facebook!

OAA is up and running on Facebook! OAA has a "Group" and also a "Cause" page for collecting donations and creating awareness. Because of security, the OAA "Group" page is only open to OAA Familes and Professionals. However, OAA's "Cause" page can be shared with your friends! Visit the OAA Cause page and click the "share" button in the upper right-hand corner. Then, enter your friend's names and send them a link to our page! The OAA Facebook Group and Cause is another great way to connect to friends and create awareness for our organic acidemias!

Marisa Cotrina Runs for OAA and PAF in the New York Marathon!

Marisa Cotrina, mom to Gabriel, Propionic Acidemia planned the most awesome feat – to run in the New York Marathon – below is Marisa' s account of the day:

I had to wake up at 5:00 am to be at the Ferry at 6. Everything closed at 7:30 am and had to wait with all the runners for 3.5 hours until my start time. Really, really cold, no tents, no nothing. Luckily, I had been told to be prepared for the cold (43 degrees) and wore 3 layers of disposable. Everybody wears clothes that you then leave at the start. Volunteers collect them all and give them to charity. People carried even sleeping bags, blankets or whatever to feel warm.

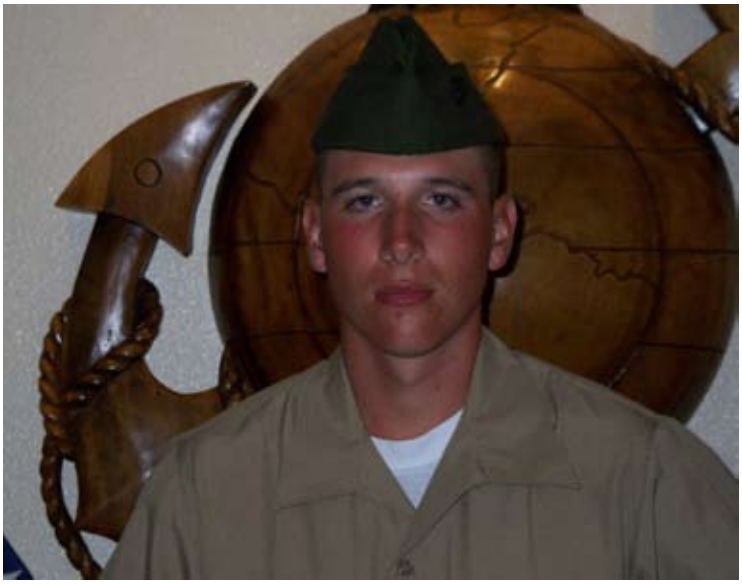
The race: FANTASTIC!!! What an experience. I had the greatest time. The public was awesome, everybody cheering and feeling for you. Children line along the race course to give you high-five, bananas, tissue, candy,... whatever they thought it might help. I had so much fun!!!

Statistics:
TIME: 4:26:01 (all in one piece!)
PLACE: 20932 out of 38356
By gender: 5274 out of 13002 women
By age: 997 out of 7069

Fundraising:
We collected $9,366 for Propionic Acidemia Foundation and $565 for Organic Acidemia Association.

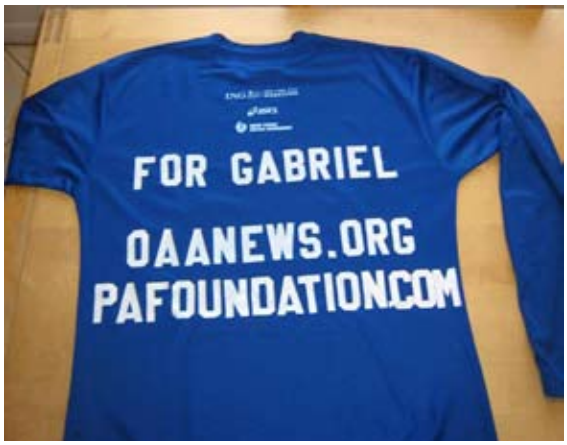
THANK YOU all for your inspiration. Thanks also to all the families that contributed. Really, it meant a lot to us. We were thrilled on how everything work out and it was an amazing experience.
I am sending a couple of picture – one of me taken during the run and my "special" shirt for the occasion!

Brandon Pescherine, IVA, a new Marine!



Brandon Pescherine is probably the first IVA patient to graduate Marine bootcamp! He successfully survived "The Crucible," and his mom Norma reports, "We couldn't be prouder of our new Marine and we feel so blessed he did so well! Before he left in July, he told me he hoped to be the first one with IVA to make it through the Marines. I don't know 100% for sure, but because he has a rare variant, he's probably right!"

This is such an awesome accomplishment for one of our 'kids'!!! Way to go Brandon!



Marisa Cotrina's "special" shirt

Michael Anthony Clapcich, MMA, Cbl C, Age 14

Michael was born on August 23, 1994. He was born right on his due date, 6 lbs. 10 ozs. A few days after birth, we went for his first newborn visit. All seemed well. He received his first shots with no side affects. Over the next day or two Michael was sleeping for longer and longer periods. Being a first time mom I thought I was very lucky to have a newborn who was already sleeping through the night. From here, our story sounds very much like the stories other OAA families have shared in previous newsletters.

Michael crashed and burned very quickly. I think in a way we were lucky that events happened the way they did. I believe Michael is functioning as well as he is today partly because he was diagnosed at two weeks old. He stopped feeding, became lethargic and wouldn't open his eyes. He was slowly going into a coma.

We were admitted into Columbia Presbyterian Babies Hospital in New York City after our pediatrician here in New Jersey couldn't tell us for sure what was wrong. At Columbia, Dr. Daryl DeVivo, a neurologist, diagnosed him with an inborn error of metabolism- an inability to metabolize B12. We stayed at Columbia for about 3-4 weeks. In that time at the hospital Michael's skin sample was sent to Dr. Rosenblatt in Canada to make a definite diagnosis and Michael was started on the treatment regime that is so very familiar to other cobalamin families.

We were home in time to celebrate his first Halloween. Getting Michael to eat was the focus of our existence for the next year and a half. At about 18 months we finally gave in and had a feeding tube placed. That was probably one of the best decisions we made. He was receiving OT, PT and speech. By two and a half he underwent strabismus surgery to correct the crossing over of both his eyes.

By this time we had started going to Johns Hopkins under the care of Dr. Ada Hamosh. (Both surgeries were done at Hopkins.) To date, we are under her care and see her twice a year for bloodwork, measure growth and to meet with a nutritionist.

Michael has a pretty cool dad and uncle. Not only are both brothers physicians, they happen to also be musically talented. As an added bonus his uncle, the drummer, did a residency at Johns Hopkins and this is how we came to know of their genetic department. He now works at Columbia Presbyterian which I mentioned above is where Michael was diagnosed. About four years ago the two brothers decided to use their music as an avenue to raise money for Michael's cause. This way the beginning of The Michael Clapcich Fund for Retinal Research. We started a website for Michael which many of you may have already looked at:www.michaelsfund.org and we are a registered- nonprofit 501©3 corporation.

We try to have one large fundraiser yearly and a few smaller ones throughout the year. This year we had the pleasure of having Dr. Venditti come to the fundraiser we held on November 2008 in NYC. All proceeds will be directed towards Dr. Venditti's research. We met Dr. Venditti for the first time in the summer of 2006. Michael participated in the clinical study at the NIH. We hope to return for a second visit this coming April.

Today Michael is a happy 14 year old. He is a great kid who is kind, friendly, loves school, music, and listening to books on tape. He doesn't let anything stand in his way. He is currently in 8th grade. Socially and cognitively he functions at about the level of a 5th or 6th grader. He is visually impaired and receives services from the Commission for the Blind of NJ. He is learning braille in the event that he may need to use it one day. He is in smaller classes and has an aide for part of the day. He is a blue belt and will be in this years school play. He has an 11 year old brother and loves his 8 y.o. yellow lab. He is on the medications that are common household names to our families: B-12 shots 3x week, folic acid, betaine (4scoops 2x daily), carnitor, caltrate for his bones, and centrum. He eats whatever he wants. From day one we have never monitored his protein intake! This may come as a shock to many of you who painstakingly calculate protein intake everyday! It just goes to show how similar yet different our kids are. He has a natural aversion to meats—although he loves McDonalds cheeseburgers! Even so he still has a g-tube in place it is really just used for medications and when he has had a bad intake day I will give him prophree mixed with 8oz. of regular milk. He is small for his age: 5feet 1 inch and about 90 pounds.

There have been many challenges that we have had to face in the last 14 years and I am certain we will be faced with many more. Such is life. I try not to worry about the many things I am sure many others worry about. We just take one day at a time and enjoy the present because that is just what it is—a present! Maybe he will go to college, maybe he won't. Maybe he will get married one day, maybe he won't. That all remains to be seen. For the moment we are helping him to be the best that he can. That is pretty much our story. Thank you for letting us share it with you.

Josephine and Robert Clapcich
678 Chestnut St.
Stirling, New Jersey 07980
908-604-8518
michaelsfundorg@yahoo.com
www.michaelsfund.org

Questionnaire Survey for the clinical outcome of the biochemically mild phenotype of IVA diagnosed by newborn screening

Carolin Wiedmann, cand. med. and
Dr. Regina Ensenauer, MD, Pediatric Research Center,
Dr. von Hauner, Children's Hospital,
Ludwig-Maximilians-University Munich, Germany.
In collaboration with **Dr. Jerry Vockley,**
University of Pittsburgh, USA.

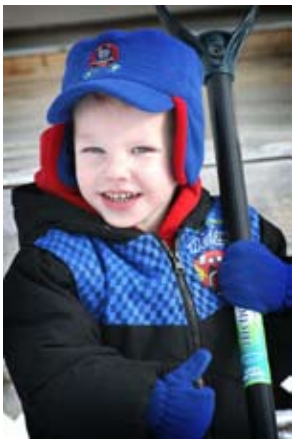
Background and research question: With the expansion of newborn screening approximately ten years ago, a novel form of isovaleric acidemia (IVA) that is associated with mild metabolic abnormalities and one particular recurring isovaleryl-CoA dehydrogenase (IVD) gene mutation, c.932C>T (p.A282V), was identified. This specific mutation was present (either on both copies of the IVD gene or on one copy of the IVD gene in combination with another mutation) in almost 70% of IVA patients of a newborn screening cohort. Therefore, the c.932C>T mutation was termed "common IVA mutation". As far as we know, affected children appear to do well. However, the clinical relevance of this mild biochemical type of IVA is still entirely unclear and the question is, whether this type confers a disease risk (at any time in life) or whether it is just a benign variant. Because of this uncertainty, treatment measures seem to vary quite dramatically: from strict dietetic leucine restriction and medication with both carnitine and glycine to nearly no treatment at all.

Study method: In order to approach this problem and consequently to facilitate both treatment of affected children and guidance of their parents, we want to initiate a retrospective study. We designed a questionnaire to learn more about all children diagnosed with the metabolically mild type of IVA by newborn screening since 1999 (when expansion of newborn screening was initiated in Bavaria, Southern Germany). Patients are eligible for inclusion in the study if molecular gene analysis has shown that the c.932C>T mutation is present in one or both copies of the IVD gene. The questionnaire mainly contains questions about the type and intensity of therapeutic measures (with regards to diet and medication), the affected child's physical and mental development, the occurrence of metabolic crises and the child's and family's quality of life. Questions are addressed to the parents. The parents are also asked to provide copies of the child's medical reports including metabolic analyses of blood (isovalerylcarnitine) and urine (isovalerylglycine). Additional studies including blood draws are not required.

Aim of the study: We expect to learn about the clinical outcome of patients with the biochemically mild type of IVA, their therapeutic management and the quality of life of affected families, given the diagnosis of a chronic disease. The results of the study are expected to be used to provide management guidelines for children with the common IVA mutation for both parents and physicians. Also, we wish to improve counselling of families with a newly diagnosed baby with the biochemically mild type of IVA.

Additional information:
If you would like to get more information, please feel free to contact us at
Carolin.Wiedmann@med.uni-muenchen.de

Gavin Brink, Isovaleric Acidemia, Age 3



June 7th 2005 was one of the best days of our life's, Gavin James was born and he was perfect! We were in the hospital for the usual few days, Gavin was a great baby and I couldn't wait to get him home and start our new life as a family. Day 3 of his life we finally got to go home, it was so great to be in the comfort of our house and get settled in, our first night went so smooth and I felt like this is what I was meant to be, a Mommy. After the first night home I was awakened by a phone call in the morning that I decided to ignore because of course I was tired and I didn't want to wake my husband up. Well a minute later my cell phone rang a couple times so I got up an listened to the message. That message changed my life and everything I thought I knew or understood about raising a child. It was a call from a pediatrician saying that Gavin's newborn screening test came back positive for something and we had to get to the hospital as soon as possible. So I called her back thinking that there had to be some kind of mistake, there was nothing wrong with Gavin, he wasn't sick, he was growing just fine and eating normal. When she confirmed that it was indeed Gavin, I lost it. How did I not notice, why wasn't I able to see that there was something wrong and would if it was too late. I knew it was serious when we got to the U of M and they were waiting at the door for us to take Gavin, that's when I got scared but realized that at least we were in the right place and hopefully we could get some answers soon as to what exactly he had and what we do from here on out.

Gavin was in the NICU at the U of M hooked up to everything you can imagine and had an IV in his soft spot. I remember they took some blood and came back to determine that he defiantly had what was called Isovaleric Acidemia. I thought I was dreaming and thought they would say this was all just a mistake but when they gave him the diagnosis of IVA the panic set in. I've never heard of such a thing and started noticing nobody has, they couldn't even pronounce it. We started doing Google searches and that didn't help, I think it just scared us more. The next day I just remember Gavin's metabolic doctor saying that he doesn't understand and that Gavin should have been sick but he showed no signs of crisis at all and his levels were fine. I don't think he had experienced a "mild" case of IVA and that is what we were told Gavin has.

He was immediately started on Glycine and Carnitine and I was able to keep breastfeeding. A few months of going that routine we ended up stopping the glycine and continuing on with the carnitine and I was able to breastfeed him for 13 months! When he started solids we didn't have to restrict or keep track of the amount of protein he was getting, I just gave him everything with less protein in it. As he got older he really seemed to self regulate his protein intake and he has ever since. Since birth he has only been really sick a few times of which I brought him to the doctor and his levels have never been high or worrisome. Right now he is our spitfire 31/2 year old, he has more energy than any other kids around and he is so smart. He has not been taking carnitine for almost a year now (except for when he's sick) and we have not noticed a change in him at all. He is also a big brother to a little brother Kaden and baby sister Raven, whom are both unaffected.

In closing I just have to say that I thank God everyday for that heel prick of blood that I truly believe saved my sons life for what it is today. Without that I don't even want to bear the idea of what would have been. Thank you all who have been such advocates for newborn screening and our children, you truly have a place in my heart because you have made a huge difference in our lives.

Callie and Brandon Brink
208 Maple St.
Chaska, MN 55318
952-448-4998
calliebrink@hotmail.com

Global Rare Disease Day Set for Feb. 28th

The Organic Acidemia Association will participate in a global Rare Disease Day on February 28th, 2009. As a Rare Disease Day Partner, we will join hundreds of other patient organizations, government agencies, medical societies and companies in focusing attention on rare diseases on that day.

This will be the Second Annual Rare Disease Day. The first was observed in Europe last year and was organized by EURORDIS, the European Rare Disease Organization, and the national alliances of rare disease patient organizations in each country.

This year, EURORDIS invited NORD, the National Organization for Rare Disorders, to organize a similar observance in the United States. Activities are also being planned in some other countries. The ultimate goal is to have a global Rare Disease Day on the last day of February each year.

The theme is that rare diseases are a public health issue, affecting millions of people around the world. The hope is that Rare Disease Day will increase awareness of rare diseases, the special challenges encountered by those affected, and the need for research to develop safe, effective treatments or cures.

As a Rare Disease Day Partner, our organization has agreed to help publicize and promote Rare Disease Day. Partners are invited to post the Rare Disease Day logo on their websites.

Also, Partners and their members are encouraged to work with their media contacts, post videos and messages online, share human interest patient stories, and nominate researchers to a Rare Disease Hall of Fame. Pins and bracelets with the Rare Disease Day logo will be available soon at cost to the members of Partner organizations.

In addition, all Partners and their members have been invited to write to their state governors to request that Feb. 28, 2009, be designated Rare Disease Day in the state. (A sample letter and resolution are posted on the NORD Rare Disease Day web page.)

Watch our website, or the following sites, for more information on these and other activities:

U.S. Rare Disease Day page on the NORD website:
http://www.rarediseases.org/rare_disease_day/rare_disease_day_info

Global Rare Disease Day website:
www.rarediseaseday.org.

Nathan Sveen, Glutaric Acidemia, Type 1, Age 3

My name is Stephanie. My husband Bruce and I are the parents of Nathan our 3 year old who has Glutaric Acidemia Type 1. Nathan was born on March 6, 2006. When I was 6 months pregnant my doctor became concerned because Nathan stopped growing, but assured us that this is common because for unknown reasons the placenta stops working. We were not concerned because I had an amniocentesis and everything was fine. I went to the hospital once a week to get non-stress tests and the baby passed every test. I had a c-section at 37 weeks, Nathan weighed 5lbs 1oz, we were so happy, he was small but everything was fine. We stayed in the hospital for 3 days and were preparing to take Nathan home when the doctor told us that Nathan's newborn screen came back and it was not normal. He said that we would need to go directly to the University of Minnesota to meet with a specialist in metabolism and genetics, he told us that Nathan would probably just need to be on "special diet". We were immediately discharged from the hospital. When we arrived at the University of Minnesota we met with Dr. Susan Berry. The first thing she said to us was, I'm so sorry to have to give you this terrible, terrible news. She explained to us that Nathan's newborn screen came back and it appears that he has Glutaric Acidemia Type 1, however sometimes the test is not correct so they wanted to do some lab work to confirm the diagnosis. I was terrified as she started to explain the disease to us. Now looking back at that moment I wasn't even listening to her, I was in shock and my entire body just went numb. I was crying harder than I have ever cried before. She assured us they would work with us and do everything they could do to try to prevent Nathan from having any negative reactions from the disease however she explained that there is no cure and no guarantees.

When we got back home the first thing I did was went on the internet and I was terrified about what I read. One week later the test came back and they confirmed the diagnosis. We were going to see Dr. Berry once a week, we met with a nutritionist we started Nathan on a special diet consisting of Glutarex 1 and Similac. They did an excellent job explaining to us how to care for Nathan. We learned that anytime Nathan gets sick with even a cold we would probably have to have him hospitalized.

The doctors suggested that if possible not to have Nathan go to a daycare due to the risk of getting sick more often. Bruce is a self employed General Contractor and I also work full-time and would not have the option of staying home with Nathan because I carry the health insurance coverage through the company that I work for. We are very fortunate to have my parents. My Mother retired about 1 year before Nathan was born and agreed to give up her retirement to watch Nathan full-time so we could work. We are very grateful to have such a wonderful support system. My Father just recently retired also. Nathan loves to go to Grandma and Grandpa's house everyday!

Besides Nathan not being a big eater the first couple of months were fine. Then he was diagnosed with acid reflux, and tortcullis which required us to start physical therapy and he was fitted for a helmet. At 3 months of age Nathan had a couple of staring spells, we seen a neurologist and Nathan was given an EEG. The Doctor explained to us that the test was "not normal" but not abnormal" and there was nothing to worry about. At 5 months of age he began having episodes where his body would jerk forward. These occurred in clusters several times per day whenever he would wake up from sleeping. We took him back to the neurologist and had a repeat EEG which revealed hysparrhythmia consistent with infantile spasms. Nathan was immediately hospitalized for four days to begin receiving ACTH steroid injections to try to stop the seizures. He was on ACTH injections for six weeks, during this time he had and increase in appetite and weight. Unfortunately he continued to have seizures throughout the ACTH therapy. At that time Keppra was started which decreased the seizures. He was having one or two clusters of seizures per day lasting five to ten minutes. We tried a couple of other seizure medications however the seizures continued. His development was delayed, he had low muscle tone and after being on the ACTH injections his appetite decreased. We started going to physical therapy once a week and eating therapy once a week.

The seizures have been our biggest battle. Currently Nathan is doing much better. His seizures have decreased. He has currently been seizure free for 7 weeks! He is on only 1 seizure medication. His development is taking off! He is a happy 3 year old with a ton of energy. We runs and plays and is a very happy little boy. He gives hugs and kisses and loves playing with our dog Buddy and our cat. He loves listening to music and rocking with Grandma. He spends a lot of time with his Grandma and Grandpa. He gets so excited when we pull up in front of their house. He loves to go for car rides. We have a cabin that we go to on weekends in the summer and he loves to play outside. He will be starting school soon so we are very excited about that.

He isn't talking yet, he goes to a speech therapist once a week. His Grandma and Grandpa take him to play group once a week at school so he is able to play with other kids. His eating is better, he only likes salty foods. He doesn't like any sweets. He is behind in his development however he continues to learn new things everyday. We are so happy and blessed with the progress he is making.

Our family has been through a lot but it has made us stronger. We were able to attend the conference in Pittsburgh this past summer. We were glad we had the opportunity to meet all of the other families and hear their stories.
We have an excellent support system, between our family and the doctors and therapist we have. We are so thankful to have Dr. Susan Berry as Nathan's metabolic doctor, she is absolutely wonderful!

Thank you for letting me share Nathan's story with you.

Stephanie and Bruce Sveen
145 9th Ave South
South St. Paul, MN 55075
651-592-5748
sveen69@comcast.net

An Overview of Nutritional Management for Organic Acidemia

Elaina Jurecki, MS, RD, Nutrition Specialist, BioMarin Pharmaceutical Inc.

General Overview of the Metabolic Pathway Dysfunction

Much has been learned about organic acidemias (OAA) over the past several years. We know that proteins are broken down into amino acids in the body which can then be utilized to make all the necessary body parts, such as skin cells, heart muscle, and liver tissue. The excess amino acids are further processed to provide energy and other important nutrients for the body. Several enzymes, including propionyl-CoA carboxylase (PC) and methylmalonyl-CoA mutase, (MM) are needed to convert four specific amino acids, isoleucine, valine, threonine, and methionine, along with other nutrients such as odd chain fats, into important compounds which can then be processed to make energy. The PC enzyme in individuals with propionic academia (PA) and the MM enzyme in methylmalonic acidemia (MMA) are not functioning appropriately, leading to a build up of several acids, with the predominant ones being propionic and methylmalonic acids. These acids are called organic acids since they have been produced in excess by the body in these disorders, and hence, PA and MMA are each termed an Organic Acidemia, or disorders leading to the excessive production and excretion of organic acids. PC and MM enzyme impairments result in the accumulation in propionic acid, methylmalonic acid, 3-hydroxy-propionic acid, methylcitric acid, along with lactic acid and ketones. The accumulation of these acids impair the Kreb cycle, which is the primary energy producing cycle in the body. The Kreb cycle takes place in the mitochondria of the cell, which is considered the energy production center. Individuals with PA and MMA have shown compromised ability to make energy in their mitochondria. Not only can the acids that accumulate in MMA and PA interfere with producing energy, but they can also interfere with the body's ability to detoxify the waste products produced from amino acids leading to the accumulation of ammonia. Cobalamin, or vitamin B12, is an important nutrient that helps many enzymes, including the MM enzyme, to function properly. Individuals with cobalamin defects will result in improperly functioning of the MM enzyme, along with the other enzymes that it helps. Consequently, you could see an accumulation of MMA in individuals with cobalamin defects as their MM enzyme will not be functioning properly due to the defect in the cobalamin function. As a result individuals with cobalamin disorders also have problems metabolizing the amino acids, isoleucine, valine, methionine, and threonine as in MMA and PA. Individuals with MMA and PA and Cobalamin defects frequently can present with frequent vomiting, lethargy, refusal to eat, and poor muscle tone resulting from the accumulation of organic acids, interference in energy metabolism, and a build up of ammonia in the body. A metabolic crisis can occur because of an inadequate calorie and/or protein intake, excessive protein ingestion, body muscle protein breakdown, and from other causes that lead to increased stressed states such as illnesses.

Similarities can be seen in other OAA's including isovaleric academia (IVA), methycrotonyl glycinuria (MCG), and glutaric aciduria type 1 (GA1), where the enzyme needed to break down an amino acid(s) to be further utilized to make energy is / are not functioning appropriately. IVA and MCG both involve malfunctioning enzymes needed to further metabolize the amino acid, leucine, to form energy. Several toxic organic acids including isovaleric acid and methylcrotonyl glycine, accumulate in the body which further compromise normal energy production. Individuals with GA1 lack an appropriately functioning glutaryl CoA dehydrogenase enzyme needed in the metabolic sequence of processing the amino acids, lysine and tryptophan, into energy. As a result, there is a toxic build up of glutaric acid and 3-hydroxy-glutaric acid in the body resulting in disruption in several metabolic processes in the body. Because of these blocks, there is a compromised ability to generate energy for the body, and an accumulation of toxic organic acids that can interfere with other body functions.

Sources of Organic Acids

Studies have been conducted to determine what the primary sources for production of the toxic organic acids are in metabolic diseases, such as MMA and PA. In one study, the investigators assessed 3 PA children and found that 50% of the PA production came from the break down of amino acids, 25% was produced by the natural bacteria found in the intestines, and the remaining 25% was assumed to be produced from the break down of body fats and proteins used to make energy during fasting. The results of this investigation suggested that the majority of the production of PA was produced from sources other than from the four amino acids received from the diet. This could explain why there is a need to implement additional treatment measures besides a dietary protein restriction for OAA's. (Thompson, Metabolism, 1990)

Goals of Therapy

Goals of therapy are to promote normal growth and intellectual development by providing an adequate nutrient and fluid intake to promote anabolism (growth) while minimizing the accumulation of toxic chemicals in the body. There are many components to treatment in addition to dietary management including ensuring adequate physical activity, receiving the necessary therapies such as physical or occupational therapy, maintaining dental health, correction of anemia, and taking the appropriate medications, such as carnitine.

Dietary Management

Dietary management will vary widely from patient to patient depending on degree of the enzyme function that each individual has and whether or not their condition presented very early when they were an infant versus later in life. It is important to tailor treatment to meet each individual's needs as one size does not fit all. Some individuals will be able to tolerate a fair amount of protein in their diet, while others will require a much more severe restriction.

Conventional dietary management of OAA's are aimed at limiting the flux through the protein degradation pathway where the malfunctioning enzyme is located, by restricting the amino acids, and with avoidance of fasting to decrease body protein and fat breakdown. These measures are important to help maintain meta-

bolic stability, but the diet also has to contain an adequate amount of calories, protein, and vitamins and minerals to ensure sufficient nutrients are received to allow for growth and development. Additional treatment measures include supplementation of carnitine to correct for deficiencies of this important nutrient. Carnitine binds to many of the toxic metabolites or organic acids that build up in the body, detoxifying them and allowing for them to be excreted in the urine. Prevention and/or aggressive treatment of infections is also important to prevent the body from producing the toxic acids due to breakdown of body proteins needed to fight the infection.

In one study, OAA patients were monitored after being fed and compared to after an overnight fast. The results showed an increased amount of toxic metabolites excreted in the urine in the fasting state as compared to the fed state. During the fasting state, the body breaks down protein stores to be utilized for an energy source. The investigators thought that during this process these proteins can not be properly utilized due to the improper functioning of the enzyme. They concluded that extended periods of fasting should be avoided in children with OAA's. (Thompson, Pediatri Res, 1990).

There have been several studies conducted to determine energy needs, or how many calories individuals with OAA's should have per day. In one study, conducted in the United Kingdom, they found a reduced rate of calories expended in patients with MMA and PA defects, 80% of that predicted based on calculations for unaffected individuals. In contrast, a report from the Netherlands showed that the calorie expenditure rate was not decreased, but slightly elevated. These investigators saw a strong correlation observed between their calorie expenditure rate and the amount of special formula consumed. This group concluded that individuals with OAA's do not have a decreased rate of calories used by the body, and that use of adequate amounts of medical formula was important to establish a normal rate of calorie expenditure. (Feillet, J Peds, 2000) (Van Hagen, JIMD, 2004). Obviously more research is needed to help understand how many calories are needed for individuals with OAA's. This is really important as offering too many calories will lead to an excessive rate of weight gain which can compromise the individual's ability to move around. Providing an inadequate amount of calories can lead to body breakdown of protein and fats to produce energy leading to an excessive production of toxic acids and metabolic instability. For now, the best approach is to work closely with the metabolic clinic to make sure a sufficient amount of calories are being provided to ensure an appropriate rate of growth and weight gain.

Metabolic Formulas

There are many metabolic formulas available for all ages and made up of different compositions. In general, these formulas provide the majority of the protein intake, from a source that limits or excludes the offending amino acids, but rich in the other amino acids. These formulas will also provide an important source of calories, vitamins, and minerals. The variety of formulas has significantly increased over the years. There are formulas specifically tailored to meet the needs of infants, and there are several formulas designed for older children and adults. These formulas are typically more concentrated in protein and not as calorically dense as compared to the infant formulas. There are additional formulas that are very concentrated in protein which can be used to treat older individuals. It is important to work closely with the metabolic dietitian to determine which is the best formula to use. Sometimes individuals with OAA's need more calories than what is provided in the formula, or can only tolerate calories without protein during illnesses. That is when a formula that only supplements calories, and possibly vitamins and minerals, come in hand. There are several of these types of formulas including Pro-Phree from Abbott Labs, PFD2 from Mead Johnson, and Duocal form Nutricia.

A study showed improved growth in OAA patients when their intakes were adequately supplemented with a metabolic formula. These individuals had their intakes optimized to promote catch up growth by increasing the calories and protein provided by the formula by 25%. Results from the study showed improvement in the growth curves for weight, increasing from 26 to 49%, length increasing from 25 to 33%, and head size increasing from 43 to 54%. The investigators also saw improvement in certain nutrient levels in their blood, which were able to reach the normal reference ranges. They concluded that by providing energy and protein for patients at recommended intakes can promote catch up growth and improve overall growth and nutritional status. (Yannicelli, Molec Genet Metab, 2003).

There was one study that looked at how compliant OAA patients were with following their diets. The investigators in this study looked at food records in terms of compliance and protein intake and found that 57% of the group over restricted their diet, taking in less natural proteins (those proteins found in foods, not from the metabolic formulas) than what was prescribed, versus 28% who liberalized their intakes taking in more than what was prescribed. Only 14% took in the equivalent amount of dietary protein prescribed. The investigators thought that the over restriction of protein was most likely due to a concern for compromising metabolic control or due to a failure to appropriately adjust the diet for growth. It is important to closely work with the metabolic clinic to make sure the diet is adequately meeting the OAA's patient's nutrient needs. (Scholl-Burgi, JIMD, 2004).

Anorexia, or loss of appetite, and vomiting are very common problems seen in children with OAA's, particularly in the first year and early in the second year of life. The reason for these symptoms is unknown, although investigators have found that some children with OAA's have unprovoked gagging during episodes of metabolic crisis. It is possible that abnormal accumulation of organic acids can affect the central regulation in the brain of the gag reflex and appetite. The placement of a gastrostomy tube greatly facilitates routine feeding and home management of minor illnesses associated with increased ketone production and times of increased caloric requirements. Some individuals with OAA's have oral motor problems which can interfere with their ability to drink, chew, and swallow. Gastrostomy tubes can help ensure that these individuals receive an adequate nutrient intake. They also greatly simplify the administration of medications. Consideration for placement of a feeding tube should be made prior to the child becoming so anorexic that it has impacted growth and development. As the child gets older, and more metabolically stable, they may no longer need a feeding tube, which can than be removed at that time.

Supplement with Essential Fats

Various fat levels were analyzed in 22 patients that had a variety of metabolic disorders in the metabolism of organic acids. The investi-

gators found that these patients, which required protein restricted diets for treatment of their metabolic disorders, had significantly lower levels of eicosapentanoic acid (EPA) and docosahexanoic acid (DHA). These are fats that are synthesized by the body from the essential fats, linoleic and linolenic acids. The investigators concluded that their diets were most likely low in essential fats since the primary dietary sources of these nutrients are typically found in higher protein foods which are excluded from the diet. (Netting, JIMD, 2002). Some patients have been found to have higher levels of triglyceride fats in their blood, due to an increased release of these fats from the liver. Other authors report that synthesis of essential fatty acids is not affected in clinically stable children with OAA's, so further studies are warranted in this area. Dietary supplementation of DHA in patients with MMA, did not have any adverse effects during the study and they were able to normalize their triglyceride levels (Aldamiz-Echevarria, JIMD, 2006). The research in the area of fat metabolism is relatively new, and there is much to be learned to determine what the optimal amounts and types of fats that should be included in the diets for individuals with OAA's.

Monitoring Metabolic Control

There are several approaches to monitor metabolic stability. Some clinics will monitor the urinary levels of metabolites, including urea, which reflects protein catabolism, along with measuring the amounts of toxic organic acids that have accumulated. Measurement of urinary ketones at home can be done on a fairly regular basis to help assess metabolic stability. An increased amount of ketones measured in the urine could signify an accumulation of toxic organic acids or that the body is not receiving enough calories resulting in the breakdown of body proteins compromising metabolic stability. Certain organic acids, such as MMA and PA, can be measured in the blood and may represent a more accurate determination of metabolic control. Clinics need to find which method will best provide useful information in determining degree of metabolic stability in their OAA patients. Besides measuring metabolic stability, there are other important parameters to monitor in OAA patients including growth and development, dietary intake, clinical exam, and measurement of other important blood components including complete blood count, measures of protein status such as albumin and prealbumin, and measures of vitamin/mineral status such as vitamin B12, folate, and iron levels.

Early and Aggressive Treatment Leads to Improved Survival and Outcome

Early detection and more aggressive treatment of babies born with OAA's are associated with improved survival rates, normalization of growth and nutritional status, and improved developmental outcomes. Metabolic stability has been observed to be easier to maintain as the OAA individual gets older. The reason may be because of less frequent illnesses and increased metabolic 'reserves' by the body which eventually can attempt to compensate for the blocked enzyme pathway, and use alternate energy sources. But many individuals continue to have problems with their mobility and cognition. Additional treatment options are being studied including organ transplant, growth hormone therapy, and antibiotic treatment, to help improve the outcome of individuals with OAA's. In the future, there may be the potential for gene therapy, which would completely correct the biochemical defect. But until then, the best approach is to work closely with the metabolic clinic to ensure the most appropriate and adequate diet is provided for management of OAA's.

References

Aldamiz-Echevarria L, P Sanjurjo, J Elorz, JA Prieto, C Perez, F Andrade, J Rodriguez-Soriano. Effect of docosahexaaenoic acid administration on plasma lipid profile and metabolic parameters of children with methylmalonic acidemia. JIMD 2006;29:58-63.

Feillet F, O Bodamer, MA Dixon, S Sequeira, JV Leonard. Resting energy expenditure in disorders of propionate metabolism. J Pediatr 2000;136:659-63.

Netting, M, D Ketterridge, M Makrides. Abnormal EFA profiles in metabolic patients on low protein diets. JIMD 2002:25 Suppl 1, 174.

Scholl-Burgi S, G Grissenauer, A Fendl, S Baumgartner, V Konstantopoulu, D Skladal. Dietary terapy in propionic academia: recommended versus real protein supply. JIMD 2004: 27(Supple 1):43.

Thompson GN, RA Chalmers. Increased urinary metabolite excretion during fasting in disorders of propionate metabolism. Pediatri Res 1990;27:413-6.

Thompson GN, JH Walter, JL Bresson et al. Sources of propionate in inborn errors of propionate metabolism. Metabolism 1990;33:1133-1137.

Van Hagen CC, EC Weber, AM van den Hurk, JH Oudshoorn, L Dorland, R Berger, T de Koning. Energy expenditure in patients with propionic and methylmalonic acidemia. JIMD 2004;27:111-112.

Yannicelli S, PB Acosta, A Velzquez, HG Block, B Marriage, TW Kurczynsl, M Miller, M Korson, RD Steiner, L Rutledge, L Bernstein, P Galvin-Parton, GL Arnold. Improved growth and nutritional status in children with methylmalonic or propionic academia fed an elemental medical food. Mol Genet Metab 2003; 80(1-2):181-8.

Colorado OA Families Meet on New Year's Eve:
The Patterson Family, The Stagni's, The Watson's and Vogt's met at Cracker Barrel to share common bonds on New Year's Eve, 2008.

OFFICIAL RECALL NOTICE

Cambrooke Foods, LLC
2 Central Street, Framingham, MA 01701
(866) 456-9776

URGENT: FOOD PRODUCT RECALL
RE: IMITATION CREAM CHEESE AND LOW PROTEIN PEANOT BUTTER™

Cambrooke Foods® is announcing a voluntary recall of all batches of the following products:

Cheddar Wizard Low Protein Imitation Cream Cheese (SKU 10310; 8 oz. jar); Herb & Garlic Low Protein Imitation Cream Cheese (SKU 10308; 8 oz. jar); and Plain Low Protein Imitation Cream Cheese (SKU 10306; 8 oz. jar).

Cambrooke Foods® is undertaking this voluntary recall as a precaution because some of these products may be contaminated with Listeria monocytogenes, an organism which can cause serious and sometimes fatal infections in young children, frail or elderly people, and others with weakened immune systems. Although healthy individuals may suffer only short-term symptoms such as high fever, severe headache, stiffness, nausea, abdominal pain and diarrhea, Listeria infection can cause miscarriages and stillbirth s among pregnant women.

For the above-listed Imitation Cream Cheese products, Listeria monocytogenes was detected through routine batch testing of product at a company facility before it was sent to customers. These batches of Imitation Cream Cheese products were destroyed. While no reports of adverse events have been reported with any product shipped to our customers, we are taking the precautionary step to recall all Imitation Cream Cheese products, regardless of batch number.

To be abundantly cautious and proactive, Cambrooke Foods® is also conducting a market withdrawal of all batches of:

Low Protein Peanot Butter™ (SKU 10809; 16 oz. jar).

This product is being withdrawn because it is produced in the same facility and using the same machinery used to produce the Imitation Cream Cheese products. Testing has not confirmed Listeria contamination in any batch of Low Protein Peanot Butter™ nor have any adverse events been reported as a result of its consumption. No other Cambrooke Foods™ product is produced using this machinery.

Cambrooke Foods™ is working directly with the Food and Drug Administration (FDA) to investigate the cause of this contamination. Cambrooke Foods™ has voluntarily suspended manufacture and distribution of these products pending its investigation.

If you have purchased any of these products since May 2008, please take the following actions immediately:

1. Discard all units of these products remaining in your possession;
2. Complete the online Recall Response Form at: http://survey.constantcontact.com/survey/a07e2f473t6fom1oqeq/start

If you have questions or require further information, contact Cambrooke Foods® toll-free 866-456-9776, ext. 1015, or via email at safetyfirst@cambrookefoods.com

Thank you in advance for your attention to this important matter. Please visit our website for regular updates.

Developing a new treatment for hyperammonemia in PA and MMA: Carbaglu

N-carbamylglutamate may decrease ammonia levels in propionic acidemia and methylmalonic acidemia patients

N-acetylglutamate (NAG) is a chemical produced in the liver that assists in removing ammonia from the body via the urea cycle. NAG deficient patients develop hyperammonemia which can be life-threatening. A person may be born with a genetic defect in N-acetylglutamate synthase (NAGS), the enzyme that produces NAG, or develop a secondary NAG deficiency such as in propionic acidemia (PA) and presumably methylmalonic acidemia (MMA). In these organic acidemias accumulation of propionyl-CoA in mitochondria of the liver decreases NAG production. Valproic acid treatment can also decrease NAG production.

Dr. Tuchman and his colleagues recently showed that a NAGS-deficient patient taking Carbaglu® (N-carbamylglutamate) had their urea cycle functions and ammonia levels restored to normal. This was done by comparing amounts of [13C] radiolabeled sodium acetate in the patient's urea before and after Carbaglu treatment. They therefore hypothesized that PA and MMA patients who develop hyperammonemia might also benefit from taking Carbaglu. To investigate this hypothesis, they treated a PA patient with Carbaglu who showed a good increase in urea production as well as a decrease in glycine glutamine and alanine levels. These results indicated that Carbaglu helped normalize the urea cycle in this patient. They now have a reliable method for measuring the effect of Carbaglu on nitrogen metabolism and the results from the patients strongly suggest that Carbaglu could be an effective treatment for either inherited or secondary NAG deficiency. Dr. Tuchman and his group would like to reproduce these results in more PA and MMA patients to see if Carbaglu can be developed into a new treatment for high ammonia in the most common organic acidemias. Hyperammonemia is only one of many possible uncontrolled metabolic factors in PA or MMA, but elimination of high ammonia could be of benefit.

NEW DRUG STUDY FOR PATIENTS WITH HYPERAMMONEMIA

If you, your child, or someone you know has a diagnosis of carbamyl phosphate synthetase I (CPSI) deficiency, N-acetylglutamate (NAGS) deficiency, Propionic Acidemia (PA), or Methylmalonic Acidemia (MMA) you or they may be eligible for this study. Eligible patients must be:

- 1 to 70 years of age
- Have a diagnosis of one of the following conditions: CPSI deficiency, NAGS deficiency, PA, or MMA
- Be willing to travel to Washington D.C. for a 4 day study (travel and lodging paid by Children's National Medical Center)

Your participation may help provide an additional treatment option for people with these conditions who suffer from elevated blood ammonia levels.

For information on cabaglu and the above studies, contact:
Mendel Tuchman, M.D.
Vice Chairman for Research
Scientific Director, Children's Research Institute
Professor of Pediatrics, Biochemistry & Molecular Biology
Children's National Medical Center
mtuchman@cnmc.org
202-476-2549 (phone)
202-476-6014 (FAX)

MEMORIALS

Noel Marie LeBlanc
MMA, Cbl C
Dec. 8, 1987 – Aug. 26, 2008

LENT FOR AWHILE

"I'll lend to you a while
A child of mine" God said,
"For you to cherish while he lives
And mourn for when he's dead.
It may be six of seven years
Or only two or three,
But, will you, till I call him Home,
Look after him for me?

He'll bring his love to gladden you
And, should his stay be brief
You'll have lots of memories
As solace for your grief.
I cannot promise he will stay
Since all from earth return,
But there are lessons taught below
I want this child to learn.

I've looked the whole world over
In my search for teachers true,
And from the throng that crowds life's lanes
At last I've chosen you.
Now will you give him all your love
Nor think your labor vain
Nor turn against me when I take
Him back to me again?"

I fancy that I hear you say,
"Dear Lord, Thy will be done,"
For all the joy this child has brought,
All fateful risks we run.

We sheltered him with tenderness,
We love him while we may,
And for the hapiness we've known,
We shall forever grateful stay.

But you came 'round to call for him
Much sooner than we'd panned-
Dear Lord, forgive this grief,
And help us understand.

Poem by Kubler-Ross, E. On Children an Death. New York: Mac-Millan Publishing Co., 1983, p. 187.

Jaden Weng
MMA,mut0, IS
Dec. 20, 2004 - Oct. 28, 2008

We are heartbroken that our son Jaden has passed away on Oct. 28th in Children's hospital at Stanford. He was in our arms when he left to a peaceful place where he will find abundance of happiness. He was also surrounded by a group of caring physicians and nurses.
About 2 weeks ago, Jaden got sick and became acidotic. After checking in the ICU, a central line was placed as Jaden is always a tough draw. Within 2 days his acidosis was quickly controlled. But then he had persistent fever and he developed sepsis. Despite excellent medical effort, Jaden's condition worsened. We were with him every minute in the last few days and he left us peacefully.

Jaden's memorable life is full of stories, fighting spirit and moments of happiness. Jaden had a severe early crisis after birth. He recovered miraculously after 7 days of comma, after aggressive life saving medical treatment. He was doing well for a couple of months until his feeding and ammonia level were off and a g-tube was required. At 7th month he had infantile spasms, one of the worst type of seizures for a child. Fortunately his seizures were resolved after half year treatment of Vigabatrin. Jaden learned to walk at 18 months, a big surprise to his physician and staffs. However Jaden's cognitive development was severely delayed. Jaden loves sound, likes to play with toys with buttons to push to make sound. Jaden is an explorer who likes to check out every room and every corner in the house.
We apologize for not updating this community for a while until now. We've been extremely busy. We've been blessed with two girls after Jaden: Joelle is 2 and 9 months and Elena is 1 year old. They have done prenatal testing and both are healthy and beautiful girls. Jaden receives excellent medical care from Lucille Packard Children's Hospital at Stanford. His regular blood test shows optimal biochemical management. Jaden is well loved by our family, the care givers, physicians, therapists, teachers, social workers and everyone who knows him. He is a special adorable boy that everyone has a special place for him.

Jaden's life has touched many people, including more than one hundred of physicians and nurses who cared for him. He taught us nothing is difficult, nothing is impossible. He also taught us happiness can be in many different forms, but the one experienced by special need child is the purest.
We wish the best to the families here. We know having a special needs child is a challenging but rewarding experience. We are so thankful to the advice and help we received from families of the Organic Acidemia Association.

Kevin and Jane Weng
Dad and Mom to Jaden, 12/20/2004 - 10/28/2008, MMA, mut0, IS
Joelle, 2 and 9 months, unaffected
Elena, 1, unaffected
Sunnyvale, CA 94087

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

Return Service Requested

NON-PROFIT ORG.
U.S. POSTAGE
PAID
MINNEAPOLIS, MN
PERMIT NO. 3395

National Organization for Rare Disorders
2008 ASSOCIATE ORGANIZATION MEMBER
NORD

sigma-tau
PHARMACEUTICALS, INC.

Remember to wear your Newborn Screening Pin to create awareness

www.oaanews.org

ANNUAL DONATION
CHANGE OF ADDRESS

Please accept $_____ as our annual tax deductible donation to the Organic Acidemia Association.

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

Name:_____

Address:_____

City/State/Zip:_____

Email Address:_____

____ I'm including $5 for a family roster

Mail to:

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

To My OAA Family:

After over 10 year of serving as the Executive Director of the OAA, I have made the decision to step aside and lessen my role. I will continue to stay involved with many of the activities of OAA - and welcome the extra help of Jana Monaco and the rest of the OAA Board of Directors. Many of you may know my family and I have gone through a most horrific time recently and while OAA is very important to me, I want to focus my 'extra' time with my family and strengthing my faith in God. I am amazed at how much OAA has grown since I took it over in 1997...we started a OAA website, an internet listserv, created family conferences, pass newborn screening legislation in many states, connected hundreds of families tog ether all along the way. That was my main goal when I set out over 10 years ago, so I guess I can feel comfortable knowing that the OAA will continue in it's next 'phase' in the hands of Jana Monaco. Thank you to all the families that have touched my life over the years.

God Bless,
Kathy Stagni

Administrative Director
Organic Acidemia Association