



Organic Acidemia Association
www.oaanews.org
Non-Profit Tax ID # 48-1038050

Organic Acidemia Association Newsletter

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January 2011

Volume XXVIII, Issue 1

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Happy New Year

As I work on this newsletter, I have tears in my eyes. I always hate to hear of one of "our kids" who has been taken from us, and in this newsletter we have three. But there is good news in here too! We have two fantastic articles from Dr. Chuck Venditti about his progress in research. We are so blessed to have him (and his team) working for our children, and apparently we are not the only ones who think so. In December Dr V was honored by President Obama with the Presidential Early Career Award for Scientists and Engineers (PECASE). Last May he was awarded the 2010 Outstanding New Investigator Award, from the American Society of Gene and Cell Therapy

This year we start planning for our next 2012 summer OAA/FOD Family Conference. Since all of our previous conferences have been EAST, we are trying to go WEST. If anyone can arrange a clinic sponsorship, please email me. Keep in mind that many people are concerned about too much heat and/or altitude.

In January Kathy will be attending a PA Consensus meeting in Washington DC. This is a major accomplishment for PA's and many experts will be attending and we are very fortunate that Kathy is "our expert". In March Marisa will be attending a conference in New York City entitled: "Genetic Diseases of Children....Advancing Research & Care Conference". Jana is continuing with her activities and committees in Washington D.C. and we can't express enough how helpful she has been in advocating for our kids, nor how grateful we are for ALL her work. In February she will be participating in a NORD conference to help define medical foods for rare diseases and discuss issues related to access experienced by many patients and families.

Also in March I will be going to the Society of Inherited Metabolic Disorders meeting, to represent the OAA. We will again share a display table with the FOD and will be handing out newsletters and brochures. I find these conferences amazing- not only for the amount of information learned, but for the sharing of data that the professionals do amongst themselves. It seems like they are always trying to learn and help our kids!

Finally, if you have not sent in your voluntary membership fee, please do so. The form is on the back of every newsletter. If you wish to donate more (for conferences, postage, or research), it will be very much appreciated! We have recently received donations in memory of those who have passed on, and do wish to express our thanks. The generosity of these families is truly amazing.

Carol Barton, Executive Director, OAA

2011 OAA Calendar Available Now!!

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



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Elizabeth Anne Malys

Propionic Acidemia May 18, 1983 - November 20, 2010

I give you this one thought to keep
I am with you still, I do not sleep
I am a thousand winds that blow
I am the diamond glints on snow
I am the sunlight on ripened grain
I am the gentle Autumn rain

When you awake in the mornings' hush
I am the swift, uplifting rush
Of quiet birds in circled flight
I am the soft stars that shine at night
Do not think of me as gone --
I am with you still 0 in each new dawn.

Elizabeth Anne Malys, 27, of Fairmount, died November 20, 2010 from complications of propionic acidemia, a condition she lived with all her life. Elizabeth brought sunshine and joy wherever she went and to everyone who knew and loved her, especially her family and friends. She gave her all to everything she did, and one of her proudest accomplishments was her graduation from West Genesee High School in 2001. Liz loved swimming, golfing, bowling, casinos and her dog, Clark. She was a communicant of Holy Family Church, where she was a



Eucharistic minister and a former member of the adult choir. Elizabeth was predeceased by her brother, R.J. (Richard J. II). Her parents would like to thank her friends and staff of Onondaga ARC on Midler Avenue for filling Liz's days with happiness.

Jordan McCann

MMA, Mut 0 - June 11, 1996 – December 9, 2010



Dear OAA Family -

I don't know if you remember me or not, I'm Jordan and Jenna's mom. I pulled off of the listserv a few years back because of the number of emails, and trying to find my work ones in the midst of it all. Anyway, I'm writing to let you know that Jordan passed away last Thursday December 9 at Hopkins after a month of her fighting back in the PICU. I always kept in touch with Joann about Stephanie, and we actually went to NIH to meet them one year so I would like it if you could pass this news on. Jordan had a very happy life, fought many battles and pulled through without complaints and was the light of my life and my best friend. She was a great big sister, and taught us more than anyone else in our lives ever did or would. Her best friend set up a website memorial page for her if anyone would like to visit it. We are choosing to have no memorial service, but will have a celebration of life on her birthday in June instead. Please share any of this information with everyone on the list. I met so many people through it, and can't remember them all. Jenna was in for most of the month with her sister, and is now home, but we may be heading back to Hopkins again today. I may jump back on the list now that life will slow down a pace or two, but in the meantime, I wanted to let everyone know.

Please include some, or all of this poem, it says it all. I have attached the last picture I took of Jordan on this past Halloween. She was in the PICU for 32 days, and we let her go to sleep at 9:30pm on the 9th with Mom, Dad and Aunt Lorraine at her side. She had acute respiratory distress syndrome, and fought very hard, but it was too much. I would like to include the website for <http://www.forevermissed/jordymccann> in the article as well. Many people have visited it and we will eventually add her story there. Jordan was a very special girl. She always helped anyone she met, and wanted to please everyone. We know she is in heaven waiting for us. Dr. Venditti came to the hospital and we made arrangements for her empty shell to be taken to NIH. We are in hopes that she will continue to give help to others through him and his studies. We will be celebrating her life on her birthday in June. Her love was Japan and anime, so we will be floating lotus flower candle lanterns in her memory so she can see them from heaven.

Kathy and family

PRECIOUS CHILD

In my dreams, you are alive and well
Precious child, precious child
In my mind, I see you clear as a bell
Precious child, precious child
In my soul, there is a hole
That can never be filled
But in my heart, there is hope
'Cause you are with me still

In my heart, you live on
Always there never gone
Precious child, you left too soon
Tho' it may be true that we're apart
You will live forever... in my heart

In my plans, I was the first to leave
Precious child, precious child
But in this world, I was left here to grieve
Precious child, my precious child

In my soul, there is a hole
That can never be filled
But in my heart there is hope
And you are with me still

In my heart you live on
Always there, never gone
Precious child, you left too soon,
Tho' it may be true that we're apart
You will live forever... in my heart

God knows I want to hold you,
See you, touch you
And I know there's a heaven
And someday I will again
Please know you are not forgotten until
then

In my heart you live on
Always there never gone
Precious child, you left too soon
Tho' it may be true that we're apart
You will live forever... in my heart

Lauren June deLima (age 9)

June 14, 2001 - October 13, 2010

Propionic Acidemia, Cardiomyopathy & Autism (passed away peacefully at Canuck Place Children's Hospice from Congestive Heart Failure)



Dear Lauren,

It has been 2 months since you've been gone and we think about you every minute of the day. We miss you deeply. This Christmas season will be especially difficult as our grief and pain are still raw, but we have peace in our hearts.

On Dec. 9th we visited your niche and found a letter from a complete stranger thanking you for shedding light on him as he was contemplating the value of life. You have more than fulfilled your purpose on earth by touching so many people's lives, especially ours. You are a true angel! We thank GOD for lending you to us and giving us the 9 memory-filled years with you. We look forward to the day we get to hug you tightly and kiss you endlessly once again.

With Love,
Mommy & Daddy

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Carbaglu News

Dear participants, families, doctors and friends,

Thank you again for your participation and support of our N-Carbamylglutamate study at Children's National Medical Center. I would like to share with you some exciting news which I received from Orphan Europe, the manufacturer of Carbaglu. Orphan Europe has now found a distributor for Carbaglu in the United States, Accredo, which has available medication for distribution. As you may already know, Carbaglu was approved by the FDA in March 2010.

Carbaglu is already in First Databank, and therefore some insurance companies which use this as a basis for their formularies will cover this medication. I now know of one patient who ordered this medication successfully and without any difficulty. The script for Carbaglu can be found on this webpage: <http://www.accredo.com/referrals/c.html> (click on "Carbaglu" for the .pdf)

Thank you again for your support of our research.
Nicholas AhMew



Network PKU Conference

Denver, Colorado
April 29 to May 1, 2011

This PKU conference will feature six small classroom workshops where Johnson & Wales School of Culinary Arts Chefs will teach and lead the class in creating low protein soups, salads, entrees and desserts that the whole family can enjoy. You will not only increase your knowledge of the low protein diet, but also increase your culinary skills. This is a rare chance to attend a weekend of cooking school at this prestigious university. Those with PKU may choose to enter the "Kitchen Stadium" for a battle of culinary technique and prowess to find out whose cuisine will reign supreme in the Iron Chef America Low Protein Cooking Challenge. \$75 per person (includes all conference materials, workshops, local ground transportation, all meals, Friday evening opening reception and Saturday evening banquet.) Due to program content, all attendees must be 12 years of age or older to attend. Additional family members not attending the full conference may register to join us for the Saturday evening banquet at the Red Lion. Cost: \$25

Registration Opens February 14th!
800-605-0410

Is a Bird in the Hand Really Worth Two in a Bush?

S.3895, the Senate's seclusion/restraint bill, "Keeping All Students Safe Act," has one provision that has created significant controversy. It is the provision which allows districts to embed a seclusion/restraint plan in and individual student's individualized Education Programs (IEP), safety plan, educational plan or behavior plan.

We oppose the IEP provision. We also oppose the passage of any bill containing the IEP provision. We oppose the provision for the following reasons:

- * Seclusion and restraint are not educational practices, strategies or methodologies. At best, they are emergency interventions.
- * Seclusion and restraint plans are not behavior plans.
- * Seclusion and restraint plans are not discipline plans; they are punishment plans.
- * Placing seclusion and restraint plans into IEPs is tantamount to declaring them "programs" within the meaning of special education law.
- * As S.3895 Finding 4, states "seclusion and physical restraint are not therapeutic.
- * As S.3895 Finding 4, also states, "seclusion and physical restraint are not effective means to calm or teach children and may have an opposite effect while simultaneously decreasing a child's ability to learn."
- * The use of seclusion and restraint as educational practices has been repudiated in therapeutic institutions including hospitals, psychiatric facilities and other residential settings for people who have challenging behaviors, even though these therapeutic institutions are staffed with medical and other highly trained professionals, and even though the physical environment itself is better suited to applying seclusion and restraint than are our nation's schools.
- * A student's IEP or educational plan is not a place to insert a seclusion/restraint plan that may result in serious injury or death to the student or to the school personnel who are tasked with implementing such a plan.
- * The current IEP process has provisions designed to address student behavior challenges, including the use of functional behavior assessments (FBAs) and behavior intervention plans (BIPs). These provisions have been in place since 1997 and were strengthened by Congress in 2004. Including seclusion/restraint plans in IEPs directly imperils all of the work that Congress, schools and parents have done to encourage the use of behavior plans.
- * IEPs are the "contract" between school districts and parents that define their child's educational expectations. Seclusion/restraint plans in IEPs, like speech therapy, physical therapy, testing accommodations, assistive technology, classroom placement, extended school year, etc., create an expectation of services to be provided. With this model, districts will naturally favor the use of seclusion/restraint plan over the development of a positive behavior support plan as the preferred method for reacting to challenging behavior.
- * No effective mechanism exists for parents to challenge the inclusion of seclusion/restraint plans in a student's individual safety plan, educational plan or behavior plan. Due process mechanisms existing for IEPs are costly, cumbersome and time-

consuming and produce additional stress for already stressed out students, parents and families.

* Including seclusion/restraint plans in IEPs will increase, rather than reduce, the use of seclusion and restraint.

* Although parents are members of the IEP team and therefore would be able to participate in the decision whether or not to insert a seclusion/restraint program into the IEP, there is no provision explicitly requiring that parents participate as full members in any group or activity creating the seclusion/restraint plan itself. This also currently is true with respect to the conducting of FBAs as well as the development of a BIP. Parents will be asked to agree to seclusion/restraint plans that have been developed without them.

The points we list above are only a partial list of reasons why Our Children Left Behind (OCLB) self-advocates and parents oppose the inclusion of IEP seclusion/restraint plans in IEPs.

The IEP seclusion and restraint provision has generated controversy. Some organizations have taken the position that they can support the bill even with the IEP provision intact, because the other provisions in the bill strengthen protections for students throughout the United States, including in states that currently have no laws or policies regulating the use of seclusion/restraint.

Other organizations are taking the position that the IEP provision is fatal to the bill. They cannot accept legislation that permits the planned use of seclusion/restraint by including it in IEPs and other similar plans. OCLB supports this position.

There is no doubt that seclusion/restraint are non-therapeutic interventions that create a significant likelihood that those who are being secluded or restrained – our nation's children, often our children with significant disabilities - and those who seclude or restrain them will be hurt or killed. Seclusion/restraint are dangerous and should not be used on our nation's children.

We are sensitive to the fact the S.3895 provides protection throughout the United States. If not for the IEP provision, OCLB would strongly support this bill. But for the reasons stated above, we believe the IEP provision, creates a greater likelihood that students will be injured or killed as a direct result of the inclusion of seclusion/restraint plans in IEPs.

More importantly, this provision will provide a strong legal basis to condone and in fact promote the use of dangerous practices that - according to Congress itself - have no educational or therapeutic value. We cannot agree that the benefit of federal regulation of seclusion/restraint in our schools outweighs the potentially fatal cost of legitimizing the use of seclusion/restraint in our children's IEPs.

Compromising in order to produce a bad bill that emboldens the use of seclusion and restraint – the Bird in the Hand – is not acceptable to us. There will be no time to go back and "fix things." The damage will be done and our children will be the worse for it. Seclusion and restraint never should be sanctioned as part of educational programming.

Tricia and Calvin Luker
www.ourchildrenleftbehind.com

Maximus Daynjer B.

MMA Mut-

People joked when I was pregnant that by naming my son Max Danger, I would be in for a lot of trips to the hospital. Of course they meant for broken bones and needing stitches.

Maximus Daynjer B. was born on May 22, 2009. He was a small but healthy 5 lbs 6 oz. He was 3 weeks early but showed no signs at birth of any health problems. Maximus took to breast feeding perfectly. He was awake, aware and seemed perfect.

When Maximus was 2 days old he started showing signs of temperature instability. He would quickly drop to 95 or 96 degrees whenever taken out from under the warmer. The doctors just attributed this to his low birth weight. Then Maximus stopped nursing. He would latch on but not suck. The lack of nutrients going in and the temperature instability caused enough concern that the neonatologist decided to place Maximus in the NICU for observation. Days 4 and 5 of his life brought no improvement. A NG tube was put into place to guarantee that we could deliver food. Then we started to see the signs of residuals. 3, 4, 5, hours after a feeding his belly was still full and the milk showed no signs of digestion. Max was dropping weight quickly at this point and was down to 4lbs. He was very lethargic and stopped responding to noise or touch.

Unfortunately I had been in this very place before. In 2001, I had a daughter named Audriey. I watched over a 13 day period in the very same NICU, her waste away and die. There was no clear cause. The doctors had no explanation. So as I watched Maximus mirror Audriey I became frantic. I told the doctor that I wanted every test done that could possibly be done. The doctor told me they believed in a mother's intuition and began a series of blood tests. Within a few hours it became clear that this was not simply "low birth weight". Maximus had an ammonia level of over 900. I was told that newborn screening was taking longer than expected to get back the results. The doctor told me that he was 100% sure it was a metabolic condition, but he did not know yet if it was compatible with life. My heart dropped.

When Maximus was 5 days old he slipped into a coma. The neonatologist consulted genetics doctors in Houston and New Orleans and made the choice to have Maximus airlifted to Tulane hospital in New Orleans. We were given the chance to hold him because they were not sure if he would survive the flight. As a family we stood together and said a prayer that Maximus would be strong and that we would be strong for him. The minister said amen, and the neonatologist walked up at that moment and said "I have his newborn screening, its methylmalonic acidemia. It is compatible with life". For the first time we had hope that Maximus could survive.



After arriving at Tulane hospital Maximus was taken into surgery and a dialysis catheter was put into place. At this point he had an ammonia level over a thousand, so we were told that that time was of the utmost importance. After 4 unsuccessful attempts to begin dialyzing Max, the doctors at Tulane decided that they could do nothing to help him. They told us that they had decided to transfer Max to New Orleans Children's Hospital. We were so scared. All we could think was more time wasted as he was put into an ambulance.

Upon arriving at Children's, Maximus was rushed into dialysis immediately. No wasted time, no paperwork. Within 30 minutes, he was on CVVH. When he arrived they drew another ammonia. At its peak, Maximus had an ammonia level of 1300. After an hour of dialysis they told us that his ammonia level was 133. We asked if the original 1300 was a mistake and we were told that they thought

that at first, but the lab ran it twice to be sure. When we asked how it could have changed so drastically in such a short time they told us "Some things just can't be explained".

After 3 hours on CVVH Maximus was switched to continual dialysis and transferred to the PICU. There Maximus continued to improve over the next few hours. The doctors told us that they were confident that they could turn off dialysis and remove the breathing tube. We sat down in the waiting room to eat for the first time in days. Moments later a nurse ran in and said "I need Max's mom and dad. He has taken a turn for the worst".

After turning off the dialysis and being extubated, Maximus began to bleed from his nose and mouth. A clot had formed on the dialysis catheter and blocked blood flow to his right side. The pressure caused his lungs to fill with blood. At 6 days old Maximus was a full code. They revived him, but we were told they did not know how much damage was done. The plan was to give large doses of blood thinner to break up the clot. Because of fear that the clot would break loose and be pulled into his heart or lungs, Max would need to be put in a coma to assure the clot would not be jolted free. Unfortunately the large doses of blood thinner made Max's brain bleed and his ventricles filled with blood. If we treated the blood clot we made the brain bleed worse and visa versa. There was a lot of time spent just waiting on tests, CAT scans, ECGs, EKGs, ultrasounds, and blood work. Three weeks later Max was brought out of his coma and he opened his eyes for the first time since 4 days old.

Maximus began to try to cry and move around. We were able to hold him and try to feed him. The blood in his ventricles was not breaking down very quickly though. The Neurosurgeons were called in and they confirmed that Maximus had hydrocephalus. Now every three days a ventricular tap was done to remove the extra fluid from his brain and relieve the pressure. Maximus's head would swell and he would not sleep or hold down any food. We were told that this is common for a child with hydrocephalus before they have a shunt placed. This went

on for several weeks. It came to a point where Maximus was not keeping any food down and his blood levels began to fluctuate. While our focus had been on his head, a blockage had formed in his transverse colon. Maximus began to crash. On the X-rays there was no clear picture of what was wrong. So Max was prepped for emergency exploratory surgery. 5 hours of surgery later they found and corrected the problem. But now on top of the mounting concern over his inter cranial pressure we were battling sepsis and a new ostomy. Max faired pretty well over the next few weeks recovering from the intestinal surgery. We were still having ventricular taps every few days waiting for the protein levels in the CSF to come down enough to facilitate the placement of a shunt. When Max was 2 months old they were able to place a ventriculo-atrial shunt. For once, everything went according to plan. At the beginning of August we were told that we could bring our baby home for the first time.

We lived on the opposite side of Louisiana from Children's hospital. The first few months home involved a lot of trips back and forth to New Orleans. We had shunt issues more than anything. Maximus never started feeding from a bottle and we just became accustomed to everything being covered in poop because of the ostomy. It was not glamorous or even fun. But Max was alive and growing. In December Max was brought into surgery again, but this was planned and anticipated. He had grown and thrived so well in those few months home that they were able to reverse the ostomy and put into place a g-tube. Everything went great and we were home by Christmas. Over the next several months things just became level and great. In February of this year Maximus got his first stomach virus and had to be hospitalized for 2 weeks. But other than checkups every few weeks we have not been hospitalized.

In March Max began physical and speech therapy. He had a lot of weakness in his right side due to the stroke in May 09, and was not able to control his head or arms well. At our first session of PT I didn't really have many expectations. Max was far beyond developmentally delayed. Speech therapy was more frightening than anything. It seemed all they did was gag him the whole time. But over the last several months we have watched Maximus do amazing things.

Today Maximus is 19 months old. He cannot walk yet, but he is very close. He wears braces on his legs to help with ankle support. We are confident that it is just a matter of time. Max still does not eat anything by mouth. Speech therapy is a lot slower progress than we would like. But he has proved time and time again that he should never be underestimated. For the first time ever last week, Maximus took a bite off a cookie, chewed it, and swallowed it!! That is what is so amazing about children like Max. He has taught us how to see the little things that matter. When he was at his worst, I made a promise to him and myself that until Max gave up, I wouldn't give up. Maximus is our gladiator. He is always happy, always smiling, and through the hard work and determination of lots of people, always trying to pull everything out of the cabinets. Thank you to everyone who has been there along the way.

Laci B. (Max's very proud mommy)
lacibernard@hotmail.com
DeQuincy, LA

Expanded Newborn Screening 2005 -2009 Missed Opportunities –

Over the past 5 years, it's become clear that many children who should have had expanded screening did not. By early 2005, the newborn screening group convened by the American College of Medical Genetics and HRSA's Maternal and Child Health Bureau had issued a consensus report that all children should be screened for 29 "core disorders", The American Academy of Pediatrics officially endorsed that report on May 12, 2005.

Thus, by at least mid-2005, the standard of care was clear: If not already performed by state mandate, hospitals and pediatricians at a minimum must give parents information about expanded newborn screening for the 29 core disorders. Tragically, there were many babies born after June 1, 2005, whose parents were not given this information. Many suffered severe injury due to late diagnosis and treatment -- injury that would have been prevented if the child had been properly screened.

Parents should know these children potentially have legal claims that, among other things, could pay for future medical and life care expenses. Most states have long statutes of limitations for child claims, so it is likely that there is still time to bring suit on behalf of children who were injured because they were not offered screening -- even babies born as far back as 2005.

Among the 29 core disorders are these 22 metabolic disorders:

1.3-MCC	3-Methylcrotonyl-CoA Carboxylase Deficiency
2.ASA	Argininosuccinate Aciduria
3.BKT	Beta-Ketothiolase Deficiency
4.CBL A, B	Methylmalonic Acidemia
5.CIT I	Citrullinemia Type I
6.CUD	Carnitine Uptake /Carnitine Transporter Defect
7.GA-1	Glutaric Acidemia Type 1
8.HCY	Homocystinuria
9.HMG	3-Hydroxy 3 - Methylglutaric Aciduria
10.IVA	Isovaleric Acidemia
11.LCHAD	Long-chain Hydroxyacyl-CoA Dehydrogenase
12.MCAD	Medium-chain Acyl-CoA Dehydrogenase
13.MCD	Multiple Carboxylase Deficiency
14.MSUD	Maple Syrup Urine Disease
15.MUT	Methylmalonic Acidemia
16.PKU	Phenylketonuria
17.PROP	Propionic Acidemia
18.TFP	Trifunctional Protein Deficiency
19.TYR 1	Tyrosinemia Type 1
20.VLCAD	Very long-chain Acyl CoA Dehydrogenase
21.BIO	Biotinidase Deficiency
22.GALT	Galactosemia

Two endocrine disorders also are covered:

1. CH Congenital Hypothyroidism
2. CAH Congenital Adrenal Hyperplasia

If your child suffered injury because of delay in diagnosis or treatment of one of these disorders and you were not offered expanded/supplemental newborn screening, you might consider contacting an attorney who specializes in these types of claims.

Chuck Hehmeyer (215-568-6190
cphehmeyer@raynesmccarty.com).

Kristen and Aimee

Isovaleric Acidemia ages 10 and 19



Hi I thought it was about time I told our story. My name is Liz and I have 5 children, 3 boys age 27, 21 and 20 and 2 girls age 19 and 10. Both my girls have IVA. Our story started when my youngest daughter Kristen was 6 weeks old. I had a normal pregnancy and birth and she was a good baby with no health problems that we were aware of. Then at 6 weeks she stopped breathing and was rushed back to the Women's and Children's hospital in Adelaide where she was born. After a few tests we were told that she had severe reflux which caused her to stop breathing and that we could treat it with medication to try to ease it. We were also told that her Guthrie test had come back and showed that she had a rare metabolic disorder and that she would have to have more tests and start on medication straight away. We were very scared as we had never heard of anything like this and didn't know what to do.

Kristen was admitted and was started on glycine. She had a skin biopsy done to confirm the diagnosis. The doctors, nurses and dietitians were all fantastic explaining everything over and over so we could understand everything we were getting told as it was a great deal to take in. Everything seemed to be going ok when she suddenly had her first "floppy". No one knew why she started having these episodes and after a lot of tests we were told they should stop as she got older or would turn into seizures. Luckily they did stop when she was about 4 but she would have them quite often sometimes lasting for 30 minutes until then. It was a shock for people to see her go unconscious in our arms with her eyes rolled back but we got used to it and knew it was o.k. We were in hospital for 2 weeks that first time.

When we got home we looked up her disorder and found as much information as we could to find out what we should be

doing to help her. My oldest son has chronic renal failure so we were used to having a sick child with special needs but it was still scary. We were in and out of hospital the first couple of years as even a runny nose made her very sick. Luckily my parents took over caring for my other children so that I could stay in hospital with her. God bless them.

Because it is genetic the doctors wanted to test me and my other children even though the other kids had a different father. We didn't think there would be a problem so we were really shocked when the tests came back and they showed my other daughter Aimee had the same thing. Looking back she did show signs. She went off milk at an early age and every time she ate meat she would get really bad stomach aches and would feel sick. Aimee also had a skin biopsy and was put on carnitine but has had no real crisis although she has got very sick at times. The girls love the fact that they have matching scars and they support each other and are very close.

Kristen was put on carnitine as well as glycine and that helped settle her levels. She has had many illnesses and has missed a lot of school but is doing really well. We find that winter is the worst time as she suffers from bronchial asthma a lot but we now get through most illnesses at home without being admitted to hospital now. She still gets sicker more often than most kids but we deal with it. The last 2 years she has had several bouts of bronchial asthma as well as whooping cough, swine flu and a flu virus and rino virus together with only 2 nights in hospital. She has had 4 lots of grommets put into her ears as she was having hearing problems and has had her tonsils out but she got through all surgeries with no problems. She also has an extra tube in her kidneys so has a few urine infections.

We have been through so much with Kristen but she deals with it all so well. She knows what she can eat and what she can't and it doesn't worry her. She loves cooking and making meals that we can eat together. Her brothers and sister and family love her and spoil her rotten as they know how lucky we are to have her with us and healthy most of the time. Kristen is a loving, caring, friendly little girl who loves animals and doing crafts and spending time with her family.

We are so lucky to have a great team of doctors and dietitians who have really done everything they can to help Kristen and keep her healthy. They are on call whenever we need them even if it's just a question or to check what to do when Kristen is sick. I am so lucky to have these special kids in my life. They have given me so much love and joy and make me appreciate every day. My eldest son should have needed a transplant years ago but has just become a father to a beautiful healthy little girl, Aimee is working in child care and studying for her diploma, Kristen is doing really well at school and is such a good girl and my other 2 boys William and Ben are doing well, working hard and are happy. What more can a mother ask for? Having a child with special needs is not a burden but a privilege and one that I will always be grateful of. I am also very lucky to have such great support especially from my mum and fiancé Mark. Thank you to everyone for the love and support we have had and still continue to have we love you all.....

Liz, Australia lizzybear642010@hotmail.com

Studies on the MMA Diet: Differing Approaches and Altered Whole- Body Metabolism

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Although dietary management is the main treatment for methylmalonic acidemia (MMA), as well as other organic acidurias, there are currently no established guidelines based on rigorous studies evaluating the different practices used by metabolic centers. Parents, patients and families are probably very aware that different centers have different approaches to nutritional management of MMA. But what are the practices used, and are there some approaches that are better than others? To begin to address this question, our goals were to: 1) document the spectrum of nutritional approaches used for the management of MMA by evaluating the diets of patients enrolled in our NIH study [“Clinical and Basic Investigations of Methylmalonic Acidemia and Related Disorders”, clinicaltrials.gov identifier: NCT00078078], and 2) to obtain objective measurements so we could examine the factors influencing the true caloric needs of the children with MMA when well. Our study on the diet and MMA (Reference 1) is one of the first to provide evidence-based measurements for dietitians managing patients with MMA to make more accurate predictions of the total calories that need to be prescribed and possibly, to help avoid obesity and its associated long-term side effects in this patient population.

We were quite fortunate and very appreciative to the 29 patients with isolated MMA (22 *mut*, 5 *cblA*, 2 *cblB*; 15 male, 14 females; age range 2 to 35 y) that could complete the study – thank you all for your help making this study possible! Study participants were evaluated at rest and were not experiencing an acute metabolic illness.

Our results showed that highly variable regimens are employed to treat patients with MMA. Most patients used a combination of protein restriction and medical food/formula supplementation (N=17/29); a number of patients were managed only with restricted natural protein intake (7/29), and a surprising number of patients received additional supplements of individual amino acids, specifically isoleucine and/or valine (N=5/29). This was puzzling to us, as there is no evidence supporting such supplementation and those are the amino acids identified in early studies as “toxic” to patients because they can turn into MMA in the body.

The most important measurement made was the baseline metabolic rate, or the resting energy expenditure (REE). Participants spent about 30 minutes in the metabolic cart. This is a system that uses a tent to measure inhaled oxygen and exhaled carbon dioxide over a short amount of time, about 10-15 min, while the participant is lying in bed. The measurements of gas exchange are used to calculate the amount of energy consumed in that period of time. These actual measurements were then compared to the values obtained by prediction based

on height, weight, sex and age that are used in metabolic clinics routinely to calculate an individual’s energy needs. It is known that in many circumstances these predictions for energy needs are not accurate for a variety of reasons. Underestimating needs can lead to underfeeding, that is providing fewer calories than a person needs resulting in malnutrition, catabolism and poor growth or, on the other hand, to overfeeding, resulting in fat storage and obesity if the estimate is too high.

Analysis of the patient’s growth and body composition showed that many of our patients are shorter than average for age, while they have increased weight and fat accumulation, as expressed by a higher percent fat mass compared to a reference population.

In this study, we also found that the measured energy expenditure was lower than predicted in most participants. This means that in some patients, the calories prescribed exceeded the patients’ needs, a factor that might contribute to a person being overweight. A detailed statistical analysis to explore the factors responsible for this lower than predicted REE showed that it was primarily due to the low skeletal muscle mass observed in patients with MMA, which is partly accounted for mostly by the low protein intake and the chronic renal failure that is a well-recognized feature of this disease.

In conclusion, this was one of the few studies that accurately assessed energy requirements in this prototypic organic aciduria. The large number of patients and the consistency of the measurements provide conclusive proof for the lower than predicted REE in MMA patients, providing a baseline reference value that could be used by metabolic physicians and dietitians to guide caloric supplementation. Similar studies during infections and metabolic decompensations would be required to better assess management during acute illness. Our research emphasizes the need for future investigations to study the effects of the different dietary practices used to treat patients with MMA, particularly a careful re-evaluation of the need for supplemental amino acids.

We want to sincerely thank the families for participating in our clinical protocol over these last 6 years since our first enrollment in 2004. We learn something new about MMA from every patient/family and your help has allowed us to obtain adequate numbers of patients required to draw powerful conclusions about an otherwise rare disorder of inborn error of metabolism.

Reference 1. Hauser NS, Manoli I, Graf JC, Sloan J, Venditti CP. (2010) Variable dietary management of methylmalonic acidemia: metabolic and energetic correlations. *Am J Clin Nutr.* [Epub ahead of print], Nov 3, PMID 21048060

Did you know that not all wheat flours are created equal?

Type	% protein	uses
High gluten	14 to 15	bagels, pizza
Whole wheat	14	hearth breads
Bread	12 to 13	traditional breads
All purpose	9 to 12	everyday cooking
Self rising	9 to 11	biscuits, cookies
Pastry	8 to 9	pie crusts, cookies
Cake	5 to 8	cakes with a lot of sugar

Effective Gene Therapy for Propionic Acidemia in Mice: One Organic Acidemia Helps Another

Randy J. Chandler, Nuria Carrillo-Carrasco, and Charles P. Venditti Organic Acid Research Section, National Human Genome Research Institute, National Institutes of Health, Bethesda, MD Email: venditti@mail.nih.gov

For many years, there has been speculation that propionic acidemia (PA) would be a good disorder to treat using gene therapy. However, when researchers created mice with PA (aka “knock-out” mice), the very real challenge of manipulating these animals became apparent. PA knock-out mice show a similar phenotype to the most severely affected humans and unfortunately, always die within 24-48 hours after birth. At this stage of life, the mice weigh around one gram and are the size of a tootsie-roll. Their small size, fragile medical condition and very short life-span make all experiments with the PA mice technically difficult.

Recently, we have had great success in the treatment of methylmalonic acidemia (MMA) mice with viral gene delivery (Reference 1). The MMA mice are very similar to the PA mice in that they also are severely affected and always die from their metabolic disorder in the first few days of life. We reasoned that an identical viral gene delivery approach should be effective in the PA mouse model because the metabolic defects that cause both PA and MMA lie in the same general biochemical pathway.

Viral gene delivery takes advantage of the natural life cycle of a virus, in which the virus normally infects cells, and then delivers its own genes. By using genetic engineering, it is possible to redesign viruses to deliver human genes. In our experiments, an adeno-associated virus (AAV) was designed to carry the propionyl-CoA carboxylase alpha subunit (PCCA) gene, which is defective in the mouse model of PA. Adeno-associated virus (AAV) is a small virus that naturally infects humans and some other primate species. AAV can infect both dividing and non-dividing cells, and express genes for long periods of time. The combination of efficient gene expression, ability to target tissues and direct sustained gene expression make AAV a very attractive candidate for use in gene therapy. Perhaps most importantly, AAV is **not currently known to cause disease; it is not a human pathogen.**

The AAV expressing the PCCA gene was then injected directly into the liver of newborn mice, and the animals were followed to determine if the gene therapy prevented death and lowered metabolite levels. The results we have observed are very encouraging: following AAV gene delivery of the PCCA gene, the propionic acidemia mice have survived for at least six months with a single treatment, and without treatment these mice uniformly perish within the first 48 hours after birth. The treated mice exhibited PCCA expression in the liver and had decreased levels of methylcitrate in the blood, which like propionic acid, is elevated in propionic acidemia. The decrease in methylcitrate levels indicates that a significant increase in propionyl-CoA carboxylase activity followed gene delivery. The

full details of these experiments have been published (Reference 2) and provide the first demonstration that gene therapy may be an effective treatment for PA in mice, and by extension, perhaps someday, in patients with propionic acidemia.

AAV has already shown promise in the treatment of other genetic diseases and has been safely used in numerous clinical trials. The results of the AAV treatment in the mouse model of propionic acidemia, the success of this approach in other genetic disease, and a historic safety record after AAV administration in humans have generated a lot of excitement. However, cautious optimism should be used since the translation of therapeutic success in animal models are sometimes difficult to replicate in humans. Extensive safety testing and FDA approval need to occur before any gene delivery clinical trial can occur.

Reference 1. Chandler R.J. and Venditti C.P. (2010) Long-term rescue of a lethal murine model of methylmalonic acidemia using adeno-associated viral gene therapy. *Mol Ther* 18(1): 11-16. PMID: 19861951

Reference 2. Chandler R.J., Chandrasekaran S., Carrillo-Carrasco N., Senac J.S., Hoferr S.E., Barry M.A., and Venditti C.P. (2010) Adeno-associated virus serotype 8 (AAV8) Gene Transfer Rescues a Neonatal Lethal Murine Model of Propionic Acidemia, *Human Gene Therapy*, [Epub ahead of print], Oct 15, PMID 20950151

PA Nutrition Guidelines

As you know, the Genetic Metabolic Dieticians International association has formed task groups to develop nutritional guidelines for genetic metabolic disorders. I wanted to share with you an update on where we are with developing the nutrition guidelines for our first organic acidemia, propionic acidemia. We have completed our first Delphi Survey where we asked a group of dietitians and doctors, with expertise in managing patients with inborn errors of metabolism, to complete a survey of approximately 90 questions on best practices for management of propionic acidemia. We will be reporting the results of this survey in a poster at the upcoming SIMD meeting next February at Asilomar, CA. Our next step is to set up a meeting of a group of experts in a Nominal group meeting to discuss treatment guideline approaches. We will then conduct one more Delphi survey to further clarify those areas where there is still some ambiguity on the best approaches for nutritional management. We are making steady progress and hope to have validated guidelines completed next year. Working on the first disorder will take the longest to complete as we get our process in place. We are hoping to merge these with medical practice as the next step.

I also wanted to make sure to mention that it would be best if we could work together with others who are interested in developing guidelines for PA. It would be great if any other professionals interested in this could work with us to keep the project coordinated. I also wanted to mention that our project is funded through a Human Research southeast regional collaborative grant.

Elaina Jurecki, MS, RD
GMDI and BioMarin

Beth's Taco Bar - (adaptable for the entire family)

2 cups shredded lettuce 1 cup diced tomatoes
1 cup salsa 1 cup sour cream
1 cup shredded cheddar cheese 1 cup shredded Jack cheese
1 cup refried beans 1 cup sliced olives
1 cup cooked and seasoned ground beef
1 cup guacamole (see recipe below)
1 dozen soft Tortillas (see recipe below)

Take a soft tortilla and fill as desired. For lower protein servings, use a smaller and thinner tortilla without meat, beans, yogurt or cheese. Serves 6.

Soft Tortillas (not low protein)

2 cups flour ¼ cup vegetable oil
2/3 cup water ½ tsp salt

Microwave the water for 1 min. Meanwhile mix the flour and salt. Add the hot water and oil at the same time and stir. Form into golf ball size balls and set in container in refrigerator to rest for ½ hr. Press or roll into tortillas and grill in a hot pan just into cooked but not very brown. Makes 12 @ 2 gm protein ea.

Guacamole

2 large ripe Haas avocados
8 oz cream cheese (read labels to get the one with less protein)
1 cup salsa
Juice of 1 lime

Soften the cream cheese in the microwave for 1 minute. Meanwhile cut open the avocados and squeeze the skins by hand until all the "guts" come out. Remove pit, add lime juice and toss. Mix in salsa and softened cream cheese.

Crista's Raisin Cake

From <http://depts.washington.edu/pku/recipes/index.html>

Cake

1/2 cup raisins 2 cups water
1/2 cup margarine 1 teaspoon vanilla
1 cup sugar 1 teaspoon cinnamon
2 tablespoon Metamucil powder 1/2 teaspoon cloves
1/4 teaspoon nutmeg 1 3/4 cup wheat starch
1 teaspoon baking soda 1 teaspoon baking powder
1/2 teaspoon salt

Boil raisins and water for 10 minutes. Add margarine and vanilla and boil for 3 minutes. In a separate bowl, combine cry ingredients. Pour raisin mixture into dry ingredients and beat with a wire whisk. Bake at 350° for 30 minutes in a greased 11" x 7" pan.

Glaze:

1 cup powdered sugar 1/4 tsp vanilla
1 tbs water

Meanwhile, whisk together glaze ingredients. Once cake has finished baking, let cool and glaze.

Per recipe: 1 g protein
Per serving: trace protein

Strawberry-Orange Ice

From <http://depts.washington.edu/pku/recipes/index.html>

1 3/4 cups orange juice 1/2 cup lemon juice
3 pints strawberries, hulled 1 3/4 cups sugar
1/8 tsp salt

In covered blender, blend all ingredients at high speed until smooth. Pour mixture into a 13x9-inch baking pan. Cover with foil or plastic wrap and freeze until partially frozen (about 4 hours), stirring occasionally. Spoon strawberry mixture into a large chilled bowl and beat with a mixer until smooth but still frozen. Return mixture to pan and freeze until firm, about 3 hours. For easier scooping and serving, remove mixture from freezer and let stand at room temperature for 10 minutes.

Makes 20, 1/2 cup servings.

Per recipe: 8.6 g protein

Per serving: 0.4 g protein

Birthday Cake with Butter Frosting

From <http://depts.washington.edu/pku/recipes/index.html>

Cake:

2 1/2 cups wheat starch 1 1/3 cups sugar
2/3 cup cake flour 4 teaspoon baking powder
1 pkg instant lemon pudding mix 4 packed tsp egg replacer
2 cups water 1/2 cup corn oil
1/4 cup margarine, softened 1 teaspoon vanilla

Preheat oven to 325°. Grease two 8" cake pans and line with waxed paper cut to fit. Mix dry ingredients, except egg replacer and sugar, in a large bowl. Whip egg replacer and 1/2 cup water until airy. Add oil, margarine, sugar, vanilla, and rest of water. Beat well. Add to dry ingredients and mix well, about 2 minutes. Pour batter into prepared pans and bake for 30-35 minutes. Allow cake to cool completely, remove from pan before frosting.

Per recipe: 5.7 g protein

Per serving (1/10 of cake): 0.6 g protein

Frosting:

1/4 cup margarine
2 cups confectioners' sugar
1 teaspoon vanilla
2 Tablespoon liquid Rich's Richwhip Topping

In a mixing bowl, thoroughly cream margarine and sugar. Stir in vanilla and Richwhip topping.

Per recipe: 0.4 g protein

Per serving: less than 0.1 g protein

Creamy Fruit Salad

From <http://depts.washington.edu/pku/recipes/index.html>

1 can fruit cocktail, drained 1 medium banana, sliced
1 apple, diced 1/2 cup seedless grapes
5 red maraschino cherries 1/4 cup kiwi, sliced
1/4 cup miniature marshmallows
1/2 cup Cool Whip non-dairy whipped topping

In a large bowl, combine fruit cocktail, sliced bananas, diced apples, grapes, cherries and marshmallows. Mix in non-dairy topping. Refrigerate at least 15 minutes before eating.

Per recipe: 4.1 g protein

Per serving (1/2 cup): 0.4 g protein



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Mail to:
Kathy Stagni
Organic Acidemia Association
13210 - 35th Avenue North
Plymouth, MN 55441

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

OAA Internet Google Group

OAA's main mission is to empower families with knowledge about organic acidemias. If you would like to connect with other families who share the same or similar diagnoses, please join our OAA Group. Visit the OAAnews.org web site to sign up. This is a private list not open to the general public (but you never know who may be "lurking").



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Facebook

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



Wanted

* Articles are ALWAYS needed for the newsletter.