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Organic Acidemia Association
NEWSLETTER

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Happy New Year 2007

As we start a new year, let's take a quick look back over the past 12 months.

The largest accomplishment was our summer conference in Dallas Texas hosted by The Institute of Metabolic Disease Baylor Health Care System. Also during the year we reported the launch of the PA Research Network (PARnet) who is focused on advancing PA studies. Our research fund trustees did another outstanding job raising money to support research that can benefit all organic academia disorders.

OAA had another opportunity to gain national visibility last year. OAA board member, Menta Pitre attended the American Society of Human Genetics annual meeting in New Orleans this past October. Thanks to the Genetic Alliance, support organizations had the opportunity to attend this scientific conference which in the past, was only open to medical professionals. Dr. Chuck Venditti and Jennifer Sloan from the NIH presented their work on MMA at this conference. Menta reports that while this conference was highly technical, it was a great opportunity to spread the word on MMA and organic acidemias. Thanks Menta for representing us!

I am also delighted to report that Board member Jana Monaco has been appointed as the Family Representative on the HRSA Advisory Committee for Heritable Disorders and Genetic Diseases in Newborn and Children. This committee makes recommendations to the Secretary of Health and Human Services with regard to the most appropriate application of universal newborn screening testing, technologies, and programs. Through her participation, the OAA gains important national visibility and continues its mission in support of newborn screening nationwide.

Looking forward, this year the OAA will produce its first "research" publication. Janice Boecker is leading this groundbreaking new project which we hope will attract the interest of researchers and medical students who would then decide to focus on our disorders. The newsletter will be written and edited by metabolic geneticists and will offer informative articles relating to each OA disorder. The plan is to publish and mail the first newsletter by the end of March.

Another exciting new development is the opportunity to collaborate with the UMDF (United Mitochondrial Disease Foundation) in the areas of research and to participate in their symposium. The UMDF has a significant amount of national exposure in the medical and scientific community and an association with them will help further OAA name/mission recognition. They are planning a scientific and family conference in Indianapolis during the summer of 2008 – so stay tuned for future info.

I would also like to take a moment to thank the OAA board of directors, who have once again provided me with the support necessary to keep the OAA working for you. I would like to introduce the newest member of the OAA board, Ruth Milne. Ruth is mom to Eilidh who has Propionic Acidemia. They live in Aberdeen Scotland and will help spread the word about the OAA to parents and professionals in Europe. Welcome Ruth.

It is now membership renewal time. If you have already sent in your renewal, we thank you for the continuing support. If not, we remind you that 100 percent of your tax deductible membership contributions are used to operate the OAA. Your contribution can be made online through our web site (www.oaanews.org) or by completing and mailing the form on the back page of this newsletter.

This year marks by TEN YEAR ANNIVERSARY leading the OAA. I would like to thank the board of directors, medical advisors, and the entire membership of the OAA for your enduring support which has enabled me to fulfill my role as leader of this organization.

Have a safe and happy new year.

Kathy Stagni, Executive Director

The Organic Acidemia Association (OAA) provides information and support to parents and professionals dealing with a set of inborn errors of metabolism collectively called 'organic acidemias'. The OAA is a volunteer organization registered with the IRS as a 501c3 non- profit corporation. Donations to the OAA are tax-deductible. OAA publishes a newsletter three times a year, hosts an internet-based listserv for information exchange and maintains a website. These services are funded by donations from corporation and individual members. Annual membership donation of $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 theopinions of our Board of Medical Advisors or Board of Directors.
- Letters and photographs sent to OAA become the property of OAAand 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.

Kristy Vos, Propionic Acidemia, Age 42

I've been meaning to write an update for a long time. I was actually waiting/hoping for something good to report. Alas, it is not meant to be. I have been diagnosed with Kidney Disease. They believe it to be Polycystic Kidney Disease. I have cysts and stones inside my kidneys, my kidneys are working at 20 percent, and my creatine level has hit 4.0. I have recently stopped working and gone on disability. I am in the process of waiting for approval from state disability. I do know they have a program for when I start dialysis. I will be getting a fistule soon in preparation for dialysis. A fistula is an access vein which they will attach to a vein in my arm. This is what they use for dialysis. My husband would like to donate a kidney to me I was just informed by our insurance company that he cannot be considered until I am on a transplant list. We are on a HMO. I have been seen by a specialist at UCLA, Dr. Stephen Cederbaum, to see if any of my problems could possibly be related to my having propionic acidemia. However, it is believed by kidney disease stems from overuse of Advil. I have always been a migraine sufferer and Advil works! Beware please because it can work to mess up your kidneys later on. My doctors have switched me to Tylenol and Maxalt for the real bad migraines. I am also taking Riboflavin for my headaches and surprisingly my headaches have lessened a bit.

I would be very interested in communicating with anyone else who has PA and kidney problems. I am on too many medications to list for you and have numerous problems along with the kidney disease, among them, I have low thyroid (I think, the one were you gain weight and become exhausted), high blood pressure, high potassium, and as previously mentioned, I am a headache sufferer.In good news, I am very thankful that my husband and I chose not to have children and I am very thankful and grateful to have a supporting husband who is willing to at least try to give me one of his kidneys. It will be a miracle if he is a match! My blood type is A Positive. In my free time, which I have a lot of now, I have written a children's book which I am trying to get published. I also enjoy spending time with our 13-year old dog, Buddy, our tortoise, Zeus, and our new kitten, Firepie.

I wish everyone the best of health,

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Chaperone Studies in Propionic Acidemia

Jan P. Kraus, Ph.D., University of Colorado at Denver and Health Sciences Center,(UCDHSC) Aurora, Colorado.

Propionic acidemia (PA) is caused by mutations in the genes PCCA or PCCB which results in a lack of the enzyme propionyl-CoA carboxylase (PCC). To be biologically functional, PCC, which is composed of alpha and beta subunits, must assemble and fold correctly inside the cell. Some mutations are known to affect the ability of the subunits to fold, and when this happens little or no functional PCC is made. One therapeutic approach to treat some PA patients, therefore, is to overcome this improper assembly and folding. Our lab is currently engaged in studying how cellular helper proteins called chaperones can restore functional PCC production in some PCCB mutations by allowing the alpha and beta subunits to aggregate and fold correctly.

Chaperones assist in polypeptide folding by unfolding forms that are non-functional, and also refolding them into functional enzyme. Our laboratory has shown that some PCCB mutations which normally produce very little enzyme can be restored to produce 30-40% functional PCC with the assistance of chaperones. We have done this by using an E.coli expression system in which mutant PCCs are able to interact with the bacterial chaperones GroEL and GroES. We are also investigating whether chemical chaperones such as betaine, valproic acid and 4-phenylbutyrate can help restore functional PCC in the presence or absence of GroEL and GroES. Our hope is to be able to develop a treatment using this approach that will clinically benefit a number of PA patients in the future.

Liz Asten, Propionic Acidemia is almost 29 now and lives in Lees Summit, MO. She has a new email address if you'd like to email her: squeak1213@yahoo.com

Brandon Harris Solomon, MMA, Cbl C, Age 11

My name is Kimberly, mother to Brandon Harris, age 11. I had a normal pregnancy until I went in for my first sonogram. There they found that I had several fibroid tumors that had caused the placenta to attach itself very low in the uterus. I was diagnosed with placenta previa. I was now a "high risk" pregnancy. I went for my normal routine appointments but continued to have several sonograms throughout the pregnancy just to make sure all was well. I continued the pregnancy to full-term without any complications.

Once Brandon was born (2 weeks late) he had to be re-hospitalized twice for billirubin. When he was born, his APGAR scores were on target. He weighed 7lbs 6 oz. and was 21 inches long. We spent the night at the hospital. Throughout the night, I tried to begin breast-feeding but Brandon did not seem at all interested in eating. We also tried bottle-feeding but the same results occurred. I had a lactation specialist come in and try to help me since this was my first child. She did help me and reassured me that this was "normal" and I was just feeling stressed and he would latch on soon. Brandon was taken the next morning for his circumcision with no complications. We were sent home once he voided. He still was not eating like I thought he should. The hospital signed me up for the lactation specialist to do some home nursing visits. Once we were home, within 24 hours, Brandon was very lethargic and looked yellow. We called the pediatrician and she wanted to see him immediately. She had lab tests done right there in her office and Brandon's billirubin levels were very high. We were admitted to St. Joseph's Hospital immediately and treated for jaundice. We were there a few days and discharged. Within 24 hours, Brandon was re-admitted for a spike in his billirubin levels. This "reoccurrence" sparked a red flag at St. Joseph's Hospital here in Baltimore because they had never had a case like this happen before. Since Brandon was not getting any better and we were not getting any answers; we asked to be sent to John's Hopkins Hospital for a complete battery of testing at the urging of a cousin who is a pharmacist. We were greeted very graciously and hastily. As soon as we got at the doors, whole teams of doctors were waiting for us. We felt like royalty at that moment. We arrived at 8:30 in the morning. At 1:30 in the afternoon, we had a partial diagnosis. Brandon was given several blood tests, head measurements, skin observations, hair swirl observations, an MRI, and CT Scans. I am sure there were many other tests completed at that time. The results concluded that he had some type of genetic disorder and was given further genetic testing. He had a fibroblast skin test taken and it was sent to a "guru" doctor in Canada. Within two weeks of his birth, was had the Cobalamin C Defect diagnosis (MMA). The doctors took Chuck and I in a conference room and began to explain this very rare disorder. We were dumbfounded and scared all at the same time. During our month-long stay at John's Hopkins Hospital NICU and PICU, Brandon did experience some seizure activity. He was given some seizure medication for a short time. The doctors concluded that the seizures were a result of Brandon's system being brought back to normal blood levels. The doctors had told us that Brandon would not have lived out that day had we not brought him there. He was a very sick little boy. We have been very fortunate that he has not had any re-occurrences since that time in the hospital. During out stay at the hospital, Brandon had to be fed through an "N-G" tube. The doctors had told us that he might need to have a stomach tube surgically placed in the future if he fails to eat. This had never had to happen. Thank goodness AGAIN! We had started on infant formula (through the consult of the nutritionist) until the doctors and MY HUSBAND urged me to be pumping my breast milk. This, we believe, has been one of the contributing factors that Brandon has done so well all of these years.

Brandon's medical regimen consists of B-12 shots 2x week, daily folic acid, Betaine (4 scoops 2x day), Carnitor, and WALNUT OIL. There is no specific evidence but Walnut Oil is said to help with the retinas and the eyes. We had nothing to lose and would try anything if it meant that Brandon would not lose his eyesight. Brandon has had no further deterioration of the retinas in several years!

We continue with periodic visits to his genetic doctor, Dr. Hamosh (about every 4-6 months), pediatric ophthalmologist, Dr. Mary Louis Collins (every 6 months), and Dr. Alan Lake (pediatrician). Dr. Collins studied under Dr. Irene

continued on page 5, Brandon Harris Solomon

Brandon Harris Solomon", continued from page 4

Maumenee who is world renowned for her work in pediatric ophthalmology studies.

Brandon loves to eat now! At times, I don't think there is enough food in the house to keep up with him. He loves to eat Wise Onion Rings, which is great because they have NO PROTEIN. I let him eat bags and bags. Dr. Hamosh always said that children with this disorder would love eating salty things. Brandon has proven this to be correct. He loves Doritos and potato chips. We calculate his protein intake EVERYDAY. He is able to eat about 40 grams of protein a day. We used to try to measure caloric intake but never reach his daily goal. I must say that I gave up trying to reach it because he continued to grow and thrive. He eats a lot of different kinds of soups (chicken noodle being the favorite) and Chef Boy R D mini meals. He loves to eat out especially at the "all-you-can-eat" buffets. He loves Chinese food (beef-n-broccoli, won-ton soup, and pork fried rice). Lo-Mein noodles are a big hit too. Brandon is very in tune with his protein levels. If he gets too much one day, his body tells him to "cut down" the next day. He also is not very hungry that day either. It is almost magical!

Brandon is a great singer and thanks to his Dad, he has been able to participate in the church choir and Sunday school programs for several years now. Chuck is the choir music director at the church. Brandon has also begun learning to play the piano just like his Dad. Brandon has been taking voice lessons as well. He has just a miraculous talent for music.

I would like to conclude Brandon's story with a simple thought…

When Brandon was born it was THE BEST day of my life! When we were dealt with this challenging hand, I felt that Brandon was given to us for a very important reason. We would rise to the occasion and put forth our best foot. I believe that he was put on this earth to one-day help others like him. I feel he will, and already is, do great things for others. Brandon is a very kind and compassionate person who loves to help in anyway he can. I am very confident that with the continued help and support of our families and friends, Brandon will continue to be a success story!

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2007 OAA Calendar Now Available at CafePress.com

http://www.cafepress.com/organicacidemia

The 2007 OAA Calendar is now ready at CafePress.com -- go check out the calendar and other items for sale at the link above.

Please remember that a portion of the sale of these items goes to support The Organic Acidemia Association.

Thank you!

Sample from inside cover of over-size 2007 calendar

Low Protein Resources

Dear Friends of
Applied Nutrition,

Thank you for helping make Network PKU 2006 a success, we couldn't have done it without you!

One of the highlights of this year's event was the workshop featuring Holiday Cooking. Chef Henkel of Johnson & Wales University did an extraordinary job making a low protein Thanksgiving menu. Please visit www.medicalfood.com and follow the link to view these and other tasty recipes.

In addition to the overview & recipes from the Network PKU conference, we have recently updated our website to include the Iron Chef America Low Protein Cooking Challenge Video and various photos taken from throughout the weekend.

To preview these items or view other Network PKU activities, please visit our website at www.medicalfood.com. For optimum viewing of the video & photos it is recommended to use Internet Explorer and a high speed connection.

Please feel free to forward or post this information to any lists, groups or individuals that may be interested.

Sincerely,
Applied Nutrition
1-800-605-0410
www.MedicalFood.com
www.DietForLife.com

Veggie Nuggets:

(whole batch there is 5.67 grams of protein from the vegetables if you use 1 potato, 1 cup carrots and 1/2 onion Some kids don't like onion so if you don't use it then subtract 1.43 grams for the onion. (total would be 4.24)

5 carrots
1 potato
1 cup zucchini

- cube veggies, add water and salt and boil until cooked
- mash
- saute onion and mix in
- drizzle olive oil over mixture
- add bread crumbs and mix in to make consistency to make a nugget
- refrigerate nuggets for approximately 30 minutes so they stay together better
- Fry

Any vegetables can be substituted in or added such as mushrooms, etc

I make a large batch and freeze them, then heat them as I need them
I started making these for Connor but my other kids eat them as well

Liz
Mom to Connor, MMA CblA

GA1 Research Golf Fundraiser

Tribune Review, Greensburg, PA
Press Release

On a clear crisp autumn morning, September 22, 2006, the International Organization for Glutaric Acidemia (IOGA) hosted their fourth golf classic in beautiful Latrobe, Pennsylvania.

IOGA, a non-profit organization serves the needs of people, mostly children, with a rare metabolic disorder. Our mission statement has not changed in ten years. The mission is to enhance the early detection and prevent neurological damage of those affected by metabolic disorders. We support research to repair brain damage and aid to rehabilitate those impaired.

We strongly support and advocate the expansion of comprehensive newborn screening for all stats. In 1995, Cay Welch attended the Pennsylvania Department of Health in Harrisburg, PA with signed petitions asking for an amendment – 'to expand the number of disorders to be screened for all 150,000 newborns in Pennsylvania.

In 1996, IOGA co-host the first International Glutaric Acidemia meeting along with The Clinic for Special Children in Strasburg, PA.IOGA advocates for families of newly diagnosed children affected by Glutaric Acidemia. Linking families to families is extremely important to the mission of the IOGA. Families are often calling to inquire about connecting with other families, specialists, and requesting information on special diets. IOGA's website (www.glutaricacidemia.org) provides open dialogue and medical abstract publications. We host medical conferences, social events, fund research and advocate the usage of stem cells.

In previous years, the IOGA golf event was privileged to have golf pro, Arnold Palmer attend, unfortunately he was attending the Ryder Cup in Ireland this year and could not attend. We are most appreciative to the many golfers who support this yearly event. Many take time out of their busy medical practices.

Among those people we wish to give special recognition are Joseph Valenzano, CEO and President of Exceptional Parent magazine. His attendance and speeches are always appreciated as he so elegantly puts a face and name to so many issues surrounding children I the special needs community.

Dr. William Zannanti, a researcher working on Glutaric Acidemia research study from Penn State, Hershey Medical Center gave a short speech on the efforts being made in advancing treatment followed by Dr. Holmes Morton, Director of the Clinic for Special Children in Strasburg, PA.

We want to specially honor Dr. Holmes Morton on his most recent award from the MacArthur Foundation, the Genius Award. IOGA has financially supported the Clinic for Special Children for many years.

We would also like to thank the Ciocca family from Walkersville, Maryland who sponsored our dinner that was enjoyed by all. There were many authentic sports memorabilia items that were added to a silent auction, plus numerous companies donated several nice door prizes. From the golf tournament, plaques and monetary prizes were awarded throughout the evening to first, second and third place winners. There were also prizes for the closest to the pin and longest drive.

Thank you also to Rob and Sharon Zygarowicz of Pittsburgh who voluntarily ran tables at registration and the door prizes among many other tasks. Volunteers are the key to our success! The Latrobe Country club was marvelous and their staffs made everything look so smooth and seamless!

Mark your calendar for next year, as IOGA plans to make the 2007 Golf Classic bigger and better than ever!

I leave you with this final thought. There is no more noble occupation in the world than to assist another human being – to help someone else succeed!

To Better Health,
Cay Welch/Mike Metil
Co-Founders of IOGA
Parents of 12 year old Michael, GA1
9638 RT 22 Highway
Blairsville, PA 15717
724-459-0179

Thank you golfers for making this such a successful event!

Christy Cranfill, MMA, Mut 0, Age 26

On December 2, 1980 after a text book perfect pregnancy and delivery our daughter was born. She had a head full of red hair and the Dr. said she was perfect. We named her Christina. We had decided on breast feeding because I had heard it was better for the baby. The second night in the hospital when they brought her to me to feed I noticed she didn't seem to want to eat. I mentioned it to the nurse and she said she may just be tired and not to worry. By morning she was in the special care nursery. She was lethargic and breathing heavy. When they took me to see her they had shaved off her red hair and had an IV in her head and a thing that looked like a keg tap where her belly button was. They gave me a picture of her that they took before they shaved her head and an envelope with the hair they had shaved. They told me they thought it was a heart problem and that they wanted to send her to Riley children's hospital.

She was transported by ambulance to Riley. It took a couple of hours for my husband to get to the hospital and get me checked out before we could join her there and by the time we got there she had been seen by the metabolic specialist at Riley. Dr. Ira Brandt. He told us that she had either methylmalonic acidemia or propionic acidemia. How they got a tentative diagnosis that fast I don't know but by finding out so fast and starting treatment immediately saved her life. Her ammonia level was 800 and her co2 was 2. I don't remember any of the other levels. It took several days to get her ammonia down and by then they had a diagnosis of MMA. She was the first MMA baby anyone at riley had ever seen much less treated. Most of what they did was done from common sense and not science. They stopped her feedings and turned up the IV to flush her system of the ammonia's and acids. We signed a form to allow them to do a kidney dialysis (spelling) but it wasn't necessary because her kidneys worked to get rid of the toxins and with the extra IV fluids she was flushed out in a few days and was able to take low protein feedings. There was no medicine for MMA. All that could be done was to put her on a protein restricted diet as her body could not process protein. I was told that I couldn't breast feed her because they needed to measure the amount of protein she took in every day and that would be impossible if she were breast fed so we switched to a mixture of Enfamil and water with vitamins and Polycose. We later replaced the water with Pedialyte because she was always vomiting and she needed the electrolytes. We also had to give her injections of b12 twice a week. When we asked what we could expect in the way of a future for her we were told not to expect her to live over a year old and that if she did live she would be brain damaged. The extent of the damage they didn't know but that there was damage. At that point I didn't care. All I wanted was for her to live. We took our baby home on Dec 19 and were admitted back into the hospital on Dec 21. We had 10 total admissions in the next 3 years. Some for a few days and some for over a month. I had 2 bags packed one had my things and one had hers. We bought night gowns and cut the shoulder seams and put ribbons on them so we could change her gowns even when she had an IV. Now days they have gowns that tie at the shoulders but back then it was a problem when you had an IV to change clothes, and with all the vomiting she did we had to change her clothes a lot.

When she was 9 weeks old they did a skin biopsy that came back proving she was not b12 receptive. That was a big disappointment but at least we didn't have to give her shots anymore. When she was 9 months old they put her on Milupa os1, a formula made in Germany that was specially made for kids that were on low protein diets. Before the Milupa she vomited several times a day. After the Milupa the vomiting doubled. It got to the point where she didn't even want to try to eat because she knew she would just be vomiting it back up. For 4 months we tried to make it work with the formula and it was the worst 4 months of our lives. After a month long stay in the hospital Dr. Wappner told us to stop the Milupa and go back to what we had been doing. She still vomited daily but not as much or as often. I know that if she had been born now instead of 25 years ago they would of installed a tube into her tummy to pump the formula in so that she would be able to take it, but 25 years ago tubing had not been thought of and by the time it was she was doing so good we didn't think it necessary.

At 2 years old she started walking and she also tossed her bottle at me and would never take it again so we went to solid foods and sippy cups. She ate things like steamed carrots and green beans and spaghettis. In fact she ate so many carrots the palms of her hands and soles of her feet turned orange which scared me half to death thinking she had a liver problem. The Dr explained that it was due to the carotene in the carrots that caused the orange color but since she loved them so much and would eat them when she wouldn't eat many other foods we got used to her orange color.

She was doing better than we ever expected her to do but she was still vomiting at least once a day and still having to go into the hospital several times a year.When she was a little over 3 years old Dr. Wappner asked us if we would be willing to try her on a research drug called carnitine. At that point I was willing to try anything that would help her so we filled out reams of forms and started on carnitine. My daughter is now 25 and has only had one hospitalization for her MMA mut0 since she started on carnitine. For us it was a miracle. When she was born they told us there were only 10 children living with MMA mut0 in the whole country. There are a whole lot more than that now. She did have some brain damage. She was dyslexic and learning to read was hard for her but we bought hooked

continued on page 9, Christy Cranfill

Christy Cranfill, continued from page 8

on phonics and she soon caught up to her class. She also had to take speech therapy for several years. She would have to work twice as hard and twice as long on her school work as other kids to get it right but she did the work and most times she ended up on the honor roll. She made up for a leaning disability with determination and hard work. The only true disability she has now as far as the brain damage goes is that she thinks and feels like a 12 year old. Her social skills are way behind and don't ask her a question if you don't want a totally honest answer. Things are black or white and there is no grey area with Christy. If she's hot she will roll her pants legs up and not care where she is or if people think she looks silly. Once in high school she walked into the bathroom where two girls were smoking and gave them a lecture on the danger of smoking. The only thing that saved her from getting beat up was that one of the girls knew about Christy and stopped the other girl from hitting her. Because of this when it came time for college Dr. Wappner said she shouldn't go to college alone so I started with her. We went for 3 semesters till I had to drop out for health reasons. If you ask Christy she will tell you we had to drop out until mom looses enough weight to get around campus. This is the truth but not how I would put it. Physically she is not as strong as others her age and she wares out faster than most. She also sleeps 12 hours a day and isn't as active as others her age. We have to be very careful of germs because any illness can trigger an acidotic episode .She has never tasted meat or nuts or drank a glass of milk. She only gets around 25 grams of protein a day and those come from mostly fruit and vegetable.

This spring we went to the N.I.H. in Maryland because they wanted to find out why Christy is doing so much better than expected. She is the oldest MMA mut0 child with her own organs. The acid that MMA mut0 kids have in their body's usually causes them to need transplants by the time they reach their 20's. The test they ran showed little or no damage to her organs. Dr Venditti has her D.N.A. and will be studying that over the next few years looking for an answer to why she is doing so well. Our greatest hope is that research will find something to give all the MMA children a chance at longer, healthier, fuller lives.

Debbie & Christy Cranfill
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Michelle Crook, MMA, Mut 0, Age 22

My name is Michelle Crook. I was born in 1984 with MMA, mut 0. I had many hospitalizations because I was acidotic a lot. I live in Michigan and my specialty doctors are in Grand Rapids and at the University of Michigan Hospital, two and a half hours away. I have an older brother, Jim, who does not have MMA. He is two years older than I. I had a little sister, Stephanie, who died from MMA at age eight. I also had a little brother, Andy, who died at age six from MMA. They both died eight years ago. Nine years ago, I received a kidney transplant and an auxiliary liver transplant. That means I have my native liver as well as an extra liver on board. The liver transplant failed. There was too much damage to the liver the doctors put in. I went home with my new kidney and was put on the list for another liver. I had to walk with a walker for awhile. My whole life I had very weak muscles because I had to restrict my protein. I vomited all the time even though I drank the low protein formula. At age five I got a feeding tube because I wasn't gaining enough weight. I had a metabolic stroke at age two and had to wear AFO's on my legs ever since. I also have some athetoid movements in my feet, arms and hands. So when I came home from the first transplant I had to work really hard to get strong again. A year later I got my second grafted liver transplant. This time recovery was very rough with lots of tests and torture. I went home in a wheelchair and a year later I could walk again. I had to work really hard in physical therapy. I cried a lot but I'm glad everyone made me work hard to walk again. I still have MMA but I don't get acidotic anymore. Because of having MMA I was delayed in my physical development, speech, fine and gross motor development and in learning. I was in special education my whole life and I learned from all the great teachers I had. I took drivers training three times but I did not pass. They said I don't react fast enough to situations on the road. Now I am twenty two. I work part time in a bakery and live at home. Sometimes I help my mom in her second grade classroom. My brother, Jim, lives in Chicago. He went to college there. My family was put through so much in my life. For three years I have been helping with the MMA research at the NIH, with Dr. Venditti. I figure I owe to my little brother and sister to help in anyway I can. The only thing that kept me going in transplant recovery for six months was thoughts of them. They died from MMA but I am determined to live. I would love to see research find a cure. Someday soon I hope to get my own apartment. I'm saving my money. I like to read, play games on the computer, go to movies, shop at the mall and listen to music. Every year my mom and I sing in a musical at church. If I can help other families with MMA, write to me or e-mail me.

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Carson Fry, Glutaric Acidemia, Type 1 Age 2

(newborn screened!)

Today is Halloween. I am busy with all of the last minute preparations of finding shoes to wear with costumes, remembering where I put the pumpkin candy holders, and hoping the weather cooperates for trick-or-treating tonight. Along with all of the fun and excitement there lingers in the back of my mind a sense of grieving and sorrow. Two years ago, Halloween was a fun and carefree holiday and then my life changed forever.

Carson Alexander was born October 27, 2004; his Grandpa Ray's birthday. He arrived with an uneventful birth, came home from the hospital and was welcomed by his sister Maddie, brother William, and Grammy. All in his first week of life we celebrated his homecoming, Halloween, then my birthday, but I will never forget the day Grammy left.

I dropped my mother off at the airport and then headed off to see our pediatrician for Carson's seven day check up. Dr. Fiona Rubenstein had become a dear friend and I couldn't wait for her to meet Carson. We were shown to the exam room and waited briefly. As Fiona entered she greeted us with her normal cheerfulness and asked me to be seated. At the time I thought she was just being considerate. I soon realized however that it was much more than that. She proceeded to tell me that Carson's newborn screen results had come back abnormally and that he needed further testing. She told me that the disorder identified was so incredibly rare that chances were it was just a false result. To be cautious however, she had him admitted to the hospital for jaundice and to keep an eye on him. We spent our first night in the hospital that night but little did we know that we would spend a majority of the next two years there as well.

Within the week Carson caught his first cold and had to be transported to Cleveland Clinic by ambulance. As he was being admitted the doctors asked tons of questions. One of the doctoral residents just happened to ask if there was anything else they should know about. I told him that Carson's newborn screen had come back abnormal and within minutes he had printed off a protocol to follow in caring for him. We still had not gotten the confirmation test results back so we didn't know if Carson really had this rare disorder but the doctor's treated him with the protocol anyway. In hindsight, I truly believe it was a factor in saving his life.

A few days later we received confirmation from our doctor, Carson does in fact have Glutaric Acidemia Type 1. I had to ask several times just how to pronounce the name let alone understand what it was. We then began a series of meetings with specialists. For the next year Carson ended up in the hospital almost every 2 to 3 weeks due to poor feeding, bronchiolitis, high fever, and cold symptoms. He had a metabolic crisis at 4 months old and mild brain injury was confirmed with MRI. It has been a long two years of over 20 hospital stays, central line and g-tube placements, three nasty line infections, fighting off normal childhood ailments like colds and respiratory infections, speech, physical, and occupation therapy. Sometimes it seems like the only thing Carson didn't get was an oil change and tire rotation.

There have been a number of things that seem to have made Carson's treatment successful and given us comfort and encouragement as we have endeavored to "live normally." We have some of the most loving family and friends one could ever hope for. Our families and our church family pray daily and have been known to drop everything and get in the car or on a plane to come to our aid. Our specialists: Dr. Shawn McCandless, Dr. Art Zinn, Dr. Douglas Kerr, Dr. Sumit Parikh, RD Judy McConnell, Dr. Tom Phelps, Dr. Maria Couthino, Dr. Walter Chwalz, RN Patti Smith, RN Kathleen Nolan and a host of other doctors and nurses at Rainbow Babies and Children Hospital and Cleveland Clinic have invested in Carson's life not just his illness. They have helped us by seeing Carson not just as a sick child but a child who happens to have an illness. My greatest joy in working with these amazingly competent people is watching them smile, talk, and enjoy Carson while treating him. God has given us so many experiences and so many people to be thankful for.

I said earlier that in the back of my mind there is grieving and sorrow over many things: the loss of dreams and plans, the loss of time with my other two children, and the loss of that sense of normalcy are just a few. It needs to be said however that these things truly are just in the back of my mind. What is most important is what resides in my daily thoughts. Where I live and strive daily is a life developing new dreams and plans, a life making the most out of the moments I have with my other two kids instead of regretting the time lost, a life of abundance instead of normalcy, but most of all just simply living life today as faithfully and joyfully as possible. Carson's life has been marked by suffering but it has been defined by love, laughing and joy. We will never look back and regret a moment of Carson's life because we know that God's plans are greater than our own. We would never trade the lessons and experiences Carson has had and will continue to learn from and the perspective and growth we have experienced as well. It has been a life well worth it.

Melissa & David Fry
13946 Chillicothe Rd.
Novelty, OH 44072
440-338-6656
fryfamily1@windstream.net

Picture: Melissa, husband David, Carson, and Madison is 6 and son William is 4.

MEMORIALS

Dr. Rebecca Sue Wappner
Born in Mansfield, OH on Feb. 25, 1944
Departed on Sep. 6, 2006 and resided in Carmel, IN.

Dr. Rebecca Sue Wappner, 62 of Carmel, IN, died Wednesday, September 6, 2006 at home. Her death followed a six-month, brave and courageous battle with cancer. Rebecca was born in Mansfield, OH on February 25, 1944. She graduated Summa Cum Laude from Ohio University in 1966 with a B.S. in Zoology. While attending O.U. she held memberships in Phi Beta Kappa, Mortar Board, Phi Kappa Phi, Alpha Lambda Delta, Sigma Xi, and Iota Sigma Pi honoraries. Becky was also a member of Sigma Kappa sorority. Dr. Wappner received her M.D. degree in 1970 from The Ohio State University. She trained in Pediatrics at Children's Hospital in Columbus, OH prior to moving to Indianapolis. She was Professor of Pediatrics and of Medical and Molecular Genetics at Indiana University School of Medicine. She continued to work until shortly before her death as Staff Pediatrician and Director of Metabolism and Genetics, Department of Pediatrics, The James Whitcomb Riley Hospital for Children. She was also Director of the Pediatric Biochemical Genetics Laboratory, the Gaucher Treatment Center, and a consultant Pediatrician to Wishard Memorial Hospital. She was a Fellow in the American Academy of Pediatrics, and the American College of Medical Genetics. Dr. Wappner was a member of the American Society of Human Genetics, American Medical Association, American Medical Women's Association, Indianapolis Medical Society, Society for Inherited Metabolic Disease, the Society for the Study of Inborn Errors of Metabolism, and the International Society for Newborn Screening. She was the Indiana Project Director for the Detection and Management of Inborn Errors of Metabolism. Her publications were numerous, including Biochemical Diagnosis of Genetic Diseases, Genetics in Primary Care & Clinical Medicine, Physician's Guide to the Laboratory Diagnosis of Metabolic Diseases, and numerous chapters in OSKI'S PEDIATRICS: PRINCIPLES AND PRACTICE. She was an active member of Soroptimist International. Dr. Wappner will be sorely missed by her family, staff, patients, their families, and everyone whose life she touched. She was an extremely caring physician who was very loved and respected. Becky will be missed for her wit, her crazy sense of humor, and her endless generosity. She exuberated an overwhelming love for her patients. Dr. Wappner was preceded in death by her father, William H. Wappner, and is survived by her mother, Helen E. Wappner; sister and brother-in-law, Diane and David Kracker; nephew and his wife, Stephen and Erika Risser. Burial services will be private. A Memorial Service and Celebration of Life will be held at The First Congregational Church, 640 Millsboro Rd, Mansfield, OH 44903 on Tuesday September 12 at 11 a.m., and at Riley Hospital for Children on Thursday, September 21 at 6:00 p.m. The family has asked that donations be made to Riley Children's Foundation Dept. of Metabolism Endowment, or to The Richland County Foundation for The Dr. Rebecca S. Wappner Memorial Scholarship Fund.

Dr. Wappner and Christy Cranfill, MMA, Mut 0

Lori Lee Smith,
Propionic Acidemia
12/20/75 - 9/6/06

I Saw You
(A Tribute to My Sister)
By
Avery Smith

I saw you today in the morning dew
As brilliant as a sea of shimmering diamonds
I shared the most amazing sunrise with you today
A million shades of red so random in their perfection
I heard you today in the laugh of my children
An enchanting melody a thousand angels strong
I walked with you today and we talked about everything
….and nothing all at once
I saw you today in the changing of the leaves
The colors of your life, the close of one season
And the ushering in of another
I sat beside a stream with you today
The peaceful flow, steady and constant
I saw you today….and you were perfect
And rest assured….I shall see you again

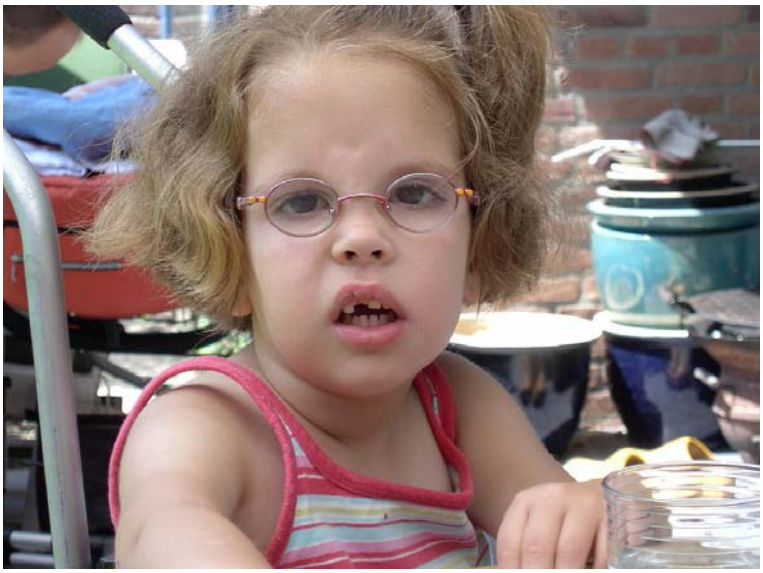
Anna Toebes-Verboven, Propionic Acidemia 8/21/1998 - 2/6/2004

On the 21st of august 1998, our beautiful girl was born. She was very small compared to her elderly sister. We were so happy en it felt so complete.Already after one day we noticed that Anna didn't wake up by herself. An easy-going baby and we both knew so well how to handle now. Breastfeeding . . . no problem! We felt very confident but not for long.

Anna couldn't maintain her own temperature well. We tried to keep her warm with aluminum foil between her blankets. Also drinking didn't go quite right. She seemed to be too tired and not really fit??!! After 2 days we couldn't cope with this situation any longer and we phoned the midwife in the evening. She assured us she would come the next morning, but 10 minutes later the door-bell rang. She happened to be around and wanted to have a look. Our heads bent over Anna's cot . . . and there the tears came. No, this wasn't good at all.Half an hour later we were in the hospital where we soon heard that Anna, our sweet little Anna, was a very sick baby. All our confidence was swept away and from this moment we stepped into a different world. The world of sick children . . .

The next day Anna was taken to another hospital, St. Radboud, a university hospital. That evening they told us the diagnose: Propionic Academia. We had never heard of metabolic diseases as a lot of other people.

Anna's first years were filled with hospital visits. In the beginning we went to St. Radboud hospital. Later, when our eldest daughter went to school, we chose for a regional hospital closer to our home. Nobody said this was good or bad, but we wanted to live a family life as normal as possible. Anna's medical treatment consisted of a glucose infusion and a schedule to build up her tube feeding. Building up her tube feeding meant decreasing the glucose. Not a lot more could be done for Anna.For us and most of all for Anna this was a burden, because finding a vein was very difficult. Besides it happened quite often that she pulled out her infusion, so she had to be injected again. I don't know how often I wanted to take my little girl with me, run away, go back home. No



more white coats, no more pain . . . just HOME. But there was no choice . . . without the hospital, she wouldn't have survived.

During the times that Anna was at home, we fully enjoyed her presence. The gap between Britt, her elder sister, and Anna became larger. It was slowly getting clear that Anna's development was different. Her physical development was impeded owing to her flabby muscles. We didn't know if she could ever walk, but when that happened at the age of 2.5 we found that a extraordinary achievement! She had managed this all by herself, who would have ever thought that this would happen!

Summer 2002, Anna's baby brother was born: Fabian. It was a miracle for us that we had a healthy son. Anna was very sweet for him and even though she couldn't talk she clearly showed she was fond of him as of her older sister.Meanwhile Anna was a regular visitor of the playgroup at rehabilitation centre De Tolbrug in Den Bosch. She really felt at home there. She enjoyed the company of the other children and the attention of her therapists. When Anna was 3 years old, she went to a school for physically handicapped children (Gabriëlschool) in Den Bosch. A special school bus picked her up and brought her home and she loved it. It became a fixed rhythm and that did her very good.

We found it therefore difficult to see that Anna every now and then got destabilized and had to go to the hospital again. Much as we did our best to keep her fit, her metabolism had often more to say. WHY did Anna

continued on page 13, Anna Toebes-Verboven

Anna Toebes-Verboven, continued from page 12

have this little error in her genes? A question you will never get an answer to, a useless question.

Summer 2003 was Anna's summer. Cadzand at the Belgian coast. Beach. Beautiful weather. Enjoy. You can read it off Anna's face. We are so complete! Britt, Anna, Fabian.

The new school year starts. Anna is throwing up a lot. It is a sign that her metabolism is not working properly. When in December it looks like Anna is going to be destabilized we immediately went to Nijmegen. We wanted to be closer to the source. Anna's diet is adjusted because she didn't grow properly the last couple of months. We have good conversations with the dietician and the metabolic doctor. Just before Christmas we are home again. Not that this means the uncertainty is over, but we are not unfamiliar with suspense and uncertainty.

January 2004 Anna breaks her leg. It costs her a great deal of energy and strength because she finds it terrible! She can't move an inch. Walking, something she loves so much because of her urge to move, isn't possible anymore. Halfway January Anna is pretty healthy despite her broken leg. It seems that her new diet is working well. That was heartening.

But then . . . on a Sunday morning, end of January. We are all having breakfast together. The tube feeding isn't finished completely before Anna throws everything up. At the end of the day Anna is in the hospital again and we are not optimistic.After 1,5 week we decide that Anna has to be transferred to St. Radboud. She doesn't seem to improve even though she often rides in her wheelchair to the playroom. But she often throws up: Anna is not fit.

On the day Anna is transferred to St. Radboud, as always I arrive in her room at 9 a.m. I discuss with the nurse . . . Anna's breathing doesn't sound as it should. I call my husband at his work and tell him that I'm feeling lonely. He decides to come this morning instead of waiting till the moment that she will be in the St. Radboud. At 10 a.m. I call him again and two words are enough: come immediately! Anna's metabolism has fallen down like a house of cards.

At noon Anna has died peacefully in my arms, she has left this world. It was enough. Anna couldn't cope with it anymore. We think she made it very clear to us to slip away so quickly. It is the 6th of February 2004, Anna is 5,5 years old.

I don't have to tell anyone what a great void Anna left behind. Her smile, her beautiful hair, her soft little hands. How can you let your child go? I don't know, but it happens. But Anna is still with us. In an other way. She will always be a part of our family. We are still with the 5 of us. Anna is our beautiful star, our flower, our princess. And she will never leave us.

Jacqueline Verboven-Toebes
19-04-2005
Drunen
The Netherlands

MEMBER MESSAGE

6th Annual Ceili for Kayleigh MMA Fundraising Event

This years 6th annual “Ceili for Kayleigh” was a huge success, raising in excess of $30,000. The event was help on Sunday November 5th, 2006 and hosted 540 people. This was the largest crowd to ever attend the annual event. We had some of the greatest prizes we have had in years including 5 professional sports jersey’s signed by that player, trips to South Beach, FL, and Wildwood, NJ, and an opportunity to sit in during the live airing of the highest rated radio show in the Philadelphia area; 610-WIP Sports Talk Radio. The most popular Irish band in the tri-state area (Pennsylvania, New Jersey, and Delaware), Blackthorn rocked the house down with most of their original music and of course the beer was flowing quite well. In addition to all the regular festivities, the Knights of Columbus Blue Gold Football Classic committee attended and presented us with a check for $4,000. Overall this was without question the best event we have had to date. All proceeds from this benefit are sent to the OAA-MMA Research fund to support the amazing work of Dr. Charles Venditti.

Marty Moran (from left to right), Andrew Young, Meghan Moran and Chuck Young

Kayleigh Moran, MMA Mut 0, with the Kitchen worker at the event, Kayleigh's teachers from her pre-school; Miss Jackie, and Miss Michelle from The Malvern School and a family friend.

Kate & Marty Moran being presented with a check from the Knights of Columbus Blue Gold All-star Classic committee.

DEPARTMENT OF HEALTH & HUMAN SERVICES Public Health Service

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October 25, 2006

Ms. Kathy Stagni
Executive Director
Organic Acidemia Association
13210 35th Avenue North
Plymouth, MN 55441

Dear Ms. Stagni:

On behalf of the National Human Genome Research Institute (NHGRI), National Institutes of Health and by the authority delegated to me through section 497 of the Public Health Service Act, as amended (42 U.S.C. 289f), I am pleased to accept the donation of $100,000.00 from the Organic Acidemia Association.

In accordance with your wishes, these funds will be used by NHGRI to support Dr. Charles Venditti's research on MMA. Support from organizations such as yours makes it possible for the National Institutes of Health to work toward understanding, preventing, and eradicating disease.

We deeply appreciate your help.

Sincerely yours,

Francis S. Collins, M.D., Ph.D.
Director

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

Return Service Requested

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 address/phone number/email address:

Name:_____

Address:_____

City/State/Zip_____

Phone Number:_____

Email Address:_____

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

Mail to:

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

OAA MEMBERSHIP RENEWAL REMINDER

A *suggested* Donation is $25 United States and $35 for International

Thank you for the many years of support that you have given to the Organic Acidemia Association. We ask that you take a moment to renew your membership today which will help us continue our valuable work providing information and support for those afflicted with organic acidemia disorders.

OAA's Services Include:

OAA Newsletter published three times/year with family stories, news for and from professionals, updates on research and much more!

Family matching—everyday OAA is matching families together for comfort and support through our internet website and family listserve.

NEW! In 2007 OAA is launching a new **OAA Research Newsletter** with informative technical articles relating to your child's disorder. We hope to attract more interest in medical students and researchers to study our disorders with this publication!

Research Funds— OAA has awarded over $150K towards research in support of improved OA treatments or a cure.

Family conferences to connect families and professionals—OAA is collaborating with the UMDF (United Mitochondrial Disease Foundation) for our next family conference. Plans are underway to join UMDF's symposium in the **summer of 2008 in Indianapolis, Indiana.**

National advocate for expanded newborn screening to help future OAA members avoid the potentially devastating effects of delayed diagnosis.

Also include the OAA info and disclaimer on this page too as in previous newsletters