Happy Summer

Summer time is finally here with its carefree, warm days and everything in bloom. I think I can speak for all that it is a welcomed season, when our kids with these inborn errors of metabolism stay pretty healthy. The opportunity to enjoy the fresh, summer air is incredibly healthy for them. It is a time of change as our children complete school and move on to the next grade level. Some of us, to include my family, get to celebrate high school graduations with one of our children and milestone birthdays!

Since January, the board of directors has been working through the transition process and acclimating to our new responsibilities and tasks, which truly take time and patience. We have held our first meeting via conference call and will have another one soon. A recent big change that we instituted was moving our list serve to a Google group. We are still working out a few minor glitches, but I think all will find this to be a much more user friendly site. It allows people to create profiles and choose discussions like that on face book, but available only to members of our group. Your patience with the change has been greatly appreciated! I am also pleased to inform you that Aimee and Leonel parents of Sapien, MMA cblA, and Marisa mother to Gabriel, PA will now be listed as Spanish translators for families who wish to connect with OAA, but are Spanish speaking only. This is a wonderful asset to our organization!

Though it seems like just yesterday that many of us met in Pittsburgh for the OAA/FOD Family Metabolic Conference, we are already in the early stages of planning for the 2010 conference. The first step is to recruit an institution that will sponsor our conference. Once we contract with a location, we can then proceed with further planning details and we will certainly keep you all posted. We are all aware of the economic challenges in general and are certainly keep you all posted. We are all aware of the economic challenges in general and are certainly feeling it on a personal level. It is my hope that despite these challenges, we will be able to fulfill our financial needs for the conference through sponsorships and fundraising. Words can not express the value of this conference for all in attendance.

The Health and Human Services Secretary’s Advisory Committee for Heritable Disorders in Newborns and Children, which I am a member of has met twice since January and continues to work diligently on newborn screening and related issues. I am happy to say that formula and the need to help make it more affordable for families is definitely a topic of concern that is being addressed. To further the importance of raising awareness for these inborn errors of metabolism as well as neurodevelopmental disabilities, I have the privilege of participating on a panel with other consumer representatives and professionals for the Intellectual Developmental Disabilities Branch of the Eunice Kennedy Shriver National Institute for Child Health and Human, to assist in reviewing the branch activities and identify its future direction. I am pleased that these topics are viewed as areas of importance and focus.

A few of our families have experienced some difficult personal losses this past season. Mr. Anthony Rouse, grandfather of Matthew Rouse was a prominent member of his Thibodaux, Louisiana community, being the founder of Rouse Markets. Though I never knew Mr. Rouse, I have come to know that his memory is felt throughout our country through his well respected business and personal affiliations, not to mention what his presence in the Rouse Family meant. His family was quite generous and thoughtful in choosing OAA and the MMA Research Fund as the beneficiaries of donations in his memory. He adored his grandson, Matthew and through that love, the $8,000 plus contributions continue to come in and will have a tremendous impact on all OAA families and Dr. Venditti’s Research Study at NIH. The Stagni family also recently lost a patriarch with the recent death of Lee’s father, Conrad Stagni, also of Thibodaux. They too chose OAA for people to make donations in his memory, because of his love for Melissa and support of the organization. These thoughtful gestures are endearing to all, because through the loss of a loved one, their memory will live on in all of us leaving a lasting impact for some very special children and educate more people on organic acidemias and the OAA. My sincere gratitude and appreciation with the deepest condolences to the Rouse and Stagni families on behalf of all!

Successful fundraisers continue to support our organization and research for these disorders. The Morans and Tates both held fundraisers in Pennsylvania raising thousands of dollars to contribute to the MMA Research Fund. The Life is a Gift Association in memory of Davide Carbone and his siblings, Just held the 2nd Annual “Walking Through Davide’s Paths”, in Italy, to raise funds for PA Research. These kinds of efforts collectively can make tremendous strides in supporting our mission.

I hope you enjoy the various articles submitted by our families on their children in this newsletter and wish you a happy, safe and healthy summer!

Jana A. Monaco
Executive Director of OAA
The Organic Acidemia Association (OAA) provides information and support to parents and professionals dealing with a set of inborn errors of metabolism collectively called ‘organic acidemias’. The OAA is a volunteer organization registered with the IRS as a 501(c)(3) non-profit corporation. Donations to the OAA are tax-deductible. OAA publishes a newsletter three times a year, hosts an internet-based list-serv for information exchange and maintains a website. These services are funded by donations from corporations 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.

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Sapien Saldivar, MMA, Cbl A, Age 2

Before giving birth to Sapien I had the most amazing, dreamy, perfect birth plan one could imagine. No interventions, no medication, Bradley method classes, hired a Doula. Well... long labor, difficult head presentation, 3 hours of pushing, an epidural, and one C-section later came our little (9lbs, 1oz) boy. All was well with him for the first 12 hours or so. Then a nurse thought he had an irregular breathing pattern, nothing to worry about, but something to observe at the NICU. He was found to have an unnamed infection and was given routine 7 days antibiotics and sent home. Fast forward to exactly 1 month before his 2nd birthday, June 30, 2008. That weekend he appeared a little tired, but nothing out of the ordinary. We even spent Saturday at a festival, swam in the river, and played. Sunday evening he was laying on the floor acting tired and we periodically noticed some irregular breathing. Since it wasn’t consistent and it was bedtime we put him to sleep and decided to call the doctor in the morning if we continued to notice anything out of the ordinary. It wasn’t until about 11am on Monday that I noticed more labored breathing and took him to the ER. After many tests they didn’t have anything definitive. Once the evening rolled around his breathing became more labored and it was decided that he would be transferred to San Antonio’s Methodist Children’s Hospital (1 hour away) for more intensive care. I know so many of you can understand the emotional rollercoaster of seeing your child unresponsive when a needle draws blood gases every 2 hours, constant IVs, doctors asking if Sapien got into any toxic chemicals (hoping this would be the case & so we could finally know the cause), neurologists explaining MRIs, and genetic doctors asking if I was a vegan, related to my husband, and did Sapien smell sweet like maple syrup. During this time Sapien had a metabolic stroke and this was apparent in his MRI. After every test imaginable they had a diagnosis on Thursday afternoon. There was incredible relief when the results from Houston arrived. It was Methylmalonic Acidemia. Methyl what? Its funny looking back and having relief as the primary emotion. When there is an incredible void of information it’s easy to grasp on to something definitive and move forward. Of course my husband and I soaked ourselves in Google searches and went on the next rollercoaster. My primary job was to get Sapien though the next 6 days of hospitalization and remain calm during the blood transfusion, learn to give him B12 injections, and laugh when the dietitian said I had to give him “this much” Propimex everyday.

Since that time Sapien has had one hospitalization (2 mos after the first) which was likely due to his decision to not eat much while battling a virus. He received fluids and antibiotics and was out in 2 days. Thank goodness for the sick day formula (double the Propimex-2 and add Polycose) which we enact each time he shows an elevated ketone level. We test him for ketones about every week when he is feeling well and several times a day when he is sick. We have a wonderful team in Austin (2 hours away) that consists of his metabolic geneticist, dietitian, social worker, and genetic counselor. The ease in communicating with them via email has been calming and allows us to touch base between visits.

Sapien’s onset was later than most. The state of Texas didn’t start their expanded newborn screening until Jan. 1, 2007, six months after his metabolic crisis. We still aren’t sure why the onset happened when it did. We theorized that since he was exclusively breastfed for so long it wasn’t until foods high in protein were introduced that his body began to show symptoms. We also wonder if his week in the NICU birth was due to his MMA, but only showing mild symptoms. From what I understand, the IV fluids and antibiotics that he received would have been the bulk of the protocol if they knew it was MMA. He did show some irregular breathing, but nurses tell me newborns go through this stage since they are so longer breathing in amniotic fluid. Perhaps we’ll never know, but I am in awe and gratitude for all the OAA parents who worked so tirelessly to have the remaining states expand their newborn screening, hoping to bypass a metabolic crisis as the avenue for finding out one has MMA.

Sapien was referred to ECI (Early Childhood Intervention) once out of the hospital. ECI had a team of a case manager, PTs, OTs, speech therapists, and a diettitian. After the initial evaluation it was decided that he needed only the case manager checking in on us. Sapien recovered amazingly well from the stroke and regained much of his muscle strength. We make sure he is often moving, climbing, and maneuvering through the playgrounds, gymnastics, and often with higher functioning kids so he can rise to their level. His muscle tone needs some strengthening, he is working on the concept of jumping- but overall he is an active and mobile toddler.

Sapien’s MMA is managed with Propimex-2, carnitine, and Bicitra. He is allowed 13 grams of protein per day. Up until last month it was always a challenge to get that much food in him. I’m beginning to rely on popcorn shrimp or ice cream at the end of the day when he needs a few more grams. He primarily eats fruits and vegetables and is beginning to like sweets. I’m sure there will come a time when I will have to start limiting his food and using low-protein products but now the issue is how to get him excited about eating anything besides McDonald’s french fries.

Sapien has adjusted much better than his parents to the MMA diagnosis. He goes on about his happy life, exploring, playing, and testing limits. We, on the other hand, are on a constant journey of letting go of expectations, inviting the diagnosis in as another family member, and allowing it to integrate into our lives. I often remind myself that Sapien’s current situation is on the better half of the MMA spectrum since he has a the Cobalamin A mutation. I also live with the knowledge that feeding tubes, hospitalizations, transplants, cognitive disabilities, and other possibilities could be on the horizon. I am grateful to our neurologist who gently shared with me this bit of wisdom in the hospital: With a healthy baby, mothers often gaze at their child thinking, but nurses tell me newborns go through this stage since they are so long breathing in amniotic fluid. Perhaps we’ll never know, but I am in awe and gratitude for all the OAA parents who worked so tirelessly to have the remaining states expand their newborn screening, hoping to bypass a metabolic crisis as the avenue for finding out one has MMA.

Aimee & Leonel Saldivar

OAA Newsletter Summer 2009
OAA Families Meet and Connect

Sage, IVA, with her mom Ann

Vince, MMA Mut 0 with his mom, Lori

The MacLean Family - Andy, MMA Mut 0

Sapien Saldivar and Zane Patterson, both MMA Cbl A, at a recent visit to the NIH

Kathy and Melissa Stagni (Propionic Acidemia)

Metabolic Family Weekend at The Hole in The Wall Gang Camp in CT. Stephanie Evans (MMA Mut 0) her sister Abby & Leah Masten (PA)

Vince, MMA Mut 0 with Sage, IVA

Vince, MMA Mut 0, Melissa, PA, Zane, MMA Cbl A, and Sage, IVA while visiting Denver at Easter

Zane Patterson, MMA Cbl A and Vince Sanchez, MMA Mut 0
Update:

Kristie Lee & Johnny
3-Methylcrotonyl-CoA
Carboxylase Deficiency, 3-MCC,
Ages 13 and 8

(Previous article written in the January 2001 OAA Newsletter)

This is an update on my children Kristie-Lee who is now 13 and Johnny who is now 8. Johnny was born Sept 20, 2000. He seemed to be a normal. We were told to make an appointment with a specialist at a Metabolic Clinic as soon as possible. We met with Dr. Paige Kaplan at Children’s Hospital of Philadelphia on Oct. 1, 2000. After 4 hours of testing and consultation, our son’s diagnosis was confirmed. We were told how fortunate we were that because of the advanced newborn screening tests this disorder was diagnosed early and if we followed a diet consisting of little or no protein, he could probably lead a normal life with-out any major complications. We were also asked to bring in our daughter, Kristie, to be tested to see if she was a carrier. On Oct. 23, we received the results of Kristie’s tests, she also tested positive for 3-MCC. This was quite a shock, as along with our son, Kristie has never showed any signs of illness, aside from the usual colds and flu.

I still didn’t understand it fully so I looked online where I found the OAA support group -- which I thought was great! I learned a lot from OAA and also met a few families whose children also had 3-MCC. Since my last update in the OAA newsletter, my daughter has never needed to be hospitalized for her 3-MCC and Johnny had one problem when he was about 3 when he got a stomach virus and needed to be kept over night at the hospital for IV fluids. Today if he still gets sick it seems to wipe him out and he takes a little bit longer then his sister to feel back to himself, but never needed any more hospital time. Luckily my children have done excellent they are on no special meds, they don’t follow any certain diets eat what they want, and we follow up at Children’s Hospital with Dr. Kaplan about once a year.

The OAA website was great when I needed someone to talk to and to learn about their disorder. Any other families whose children have 3-MCC please feel free to contact to me my email address is irish2st29@aol.com.

Connecting with OAA

OAA Listserv is Now on Google Groups!
This past spring OAA changed from our current listserv to a new “Google Group.” If you would like to connect with other families who share the same or similar diagnosis -- please join our new OAA Google Group. To sign up, visit OAA’s website at: http://www.oaanews.org/

Since this Google Group is private and requires authorization, you may be asked if you are a family member of a professional dealing with an organic academia before allowed to join.

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

OAA is on Facebook!
OAA is up and running on Facebook! OAA has a “Group” and also a “Cause” page for collecting donations and creating awareness. Because of security, the OAA “Group” page is only open to OAA Families and Professionals. However, OAA’s “Cause” page can be shared with your friends! Visit the OAA Cause page and click the “share” button in the upper right-hand corner. Then, enter your friend’s names and send them a link to our page! The OAA Facebook Group and Cause is another great way to connect to friends and create awareness for our organic acidemias!
Matthew Thomas Rouse, MMA, Cbl C, Age 19

Matthew is my fourth born child and is 19 years old. It took us 5 1/2 months of searching for the right doctor before he was finally diagnosed with Methylmalonic Aciduria, type C, with Homocystinuria. He also has hydrocephalus (which is associated with very few MMA children) and had to have a vp shunt put in to drain the fluid from his brain. The shunt is the thing that gives us the most problems. He has had to have numerous surgeries to keep it functioning. He has a heart condition also, called pulmonary stenosis. His vision is impaired but we have had a couple of surgeries to correct strabismus and nystagmus. He seems to have only peripheral vision and no depth perception. He also had something called a thyroglossal duct cyst which took two surgeries to totally remove.

Matthew eats very well, pretty much anything he wants. He takes his B-12 injection daily, along with carnitor and cystadane. He drinks pro-phree formula also. He is a very big boy, weighing in at about 165 lbs. and is 5 ft. 7 inches tall. He does walk, but will tire easily so we use a wheelchair for long distances. Plus the wheelchair gives us a little control over him. He can be very stubborn at times. He communicates to all that are with him on a regular basis. He is definitely getting more verbal everyday. The learning never stops! He is still in diapers, needs assistance to bathe, eat, dress, etc. He loves to go bowling, see movies, and play on the computer and playstation. But his favorite thing of all is to ride! He could ride around in a car with the radio playing all day long. He is absolutely fascinated with airplanes too. We take him once or twice a year to Disneyworld so he can fly and ride the monorails, buses, boats, etc.

Matthew has 3 older siblings, none are afflicted with MMA, but two are carriers. They are absolutely wonderful with their “little” brother. God knew what he was doing when he sent me Allison, Adam and Rachel first. They really do their part in playing with him and helping me when I need them. My husband, Tommy, is a big help in that he loves to play playstation as much as Matt, so that means I don’t have to learn how!

Well, I think that about covers everything. All that is left to say is that Matthew is a very complicated, exhausting, and demanding boy, but we all love him so much! He has a very sweet personality that brings out the best in everyone he meets. I truly believe that he is both a gift and a test from God. I try to do as much as I can for Matt to make him happy without spoiling him rotten. I have to admit, discipline is the one thing I have not mastered with him. My only goal is to keep him healthy and happy, happy, happy!

We would love to hear from anyone concerning MMA.

Karen and Tommy

In Memory of
Matthew’s grandfather,
Mr. Anthony Rouse,
donations were graciously sent to
the OAA/MMA Research Fund.
Archie was March 10, 2001, weighing in at 6lb 12oz and all seemed well, we had the perfect baby -- he never cried and hardly woke! At my 6 weeks check up I was told to get him to Gloucester Royal Hospital because his breathing was erratic. They informed us we were over reacting there was nothing wrong, strange as it was the doctors that picked it up not us! As he got older we realized things were not right, by 10 months this perfect baby still did nothing but lie there, now concerned we took him back to the doctors and got referred to a specialist, where we are told of his developmental delay. For months we accepted this 'diagnosis' until my husband spoke up and said “when does delay turn into concern?” At this point he was diagnosed as having “chronic Autism!” After a few months, the specialist decided there were more issues showing up, so he took more tests, and then the same test 3 more times as she could not believe the results....Then all hell broke loose!!! His bottle I was feeding him with at the time was taken from his mouth and we were told that basically we had been slowly killing our perfect son and had given him brain damage as he had the rare condition “Propionic Aciduria”...To say the least we were shocked -- what had we done to our perfect baby!!

Archie was instantly put on a low protein diet and we were referred to Bristol Children’s Hospital under Dr. Hamilton Shield, a Metabolic Specialist. Archie was put on Biotin and Carnitor (I wasn’t sure what this does, but give it him anyways as they say its best.) He has had a few “episodes” as they call it, the last one being the worse. Our hospital closes at 8pm and the next closest one refused to take him as he was so ill. Dr. Hamilton Shield called a rapid response team up from Bristol and Archie was transferred to the intensive care unit. We dread to think what would have happened otherwise. We were warned he may not make it to Bristol. But all was well in the end!

Archie is in main stream school and about 2 years behind children his age. He is a lot smaller than other children his age, looking more like a 5 year old. They told us he would not walk or talk and probably not make it to 6. Well they were wrong again -- he does not stop talking and runs around all the time. Allbeit, he gets tired quicker then most. Archie is now 8 year old, so we take what the specialists say “with a pinch of salt”. Nothing has ever really been explained to us what we know is what we found on internet. The dietician gave us a one day menu we had to call after a month and say this is not right. How can a child eat the same thing forever, everyday? My friends helped me more than the dietician and now we are protein experts!

They told us he wouldn’t live past 6, but thanks to the internet (and Facebook) again we have found Kathy (Stagni) who has a 20 year old daughter...Happy Days!

Take Care and Keep Smiling

Salli, Dave, Charlie and Archie

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**Update from the Association**

**"La Vita e un Dono” – A Propionic Acidemia Research Fund in Italy**

The association “La vita e un dono” was created with the purpose to remember Davide and his little brothers, all affected by propionic acidemia and to promote the research of therapies for this metabolic disease. Davide’s joy of living, his courage and strength in living with Propionic Acidemia, his school efforts and results remained in the heart of so many people that met him and after 2 years since he left us, everybody still remembers him and helps us with the association, which has now 175 volunteers, helping with the organization of all the events.

The Association’s activities started soon after it was founded with funding and events for the organization. An internet site was created and many articles published on local newspapers. Davide’s words are still a guide for our life - that helps the association to grow, just like the oak, seeded at the start of the fundraiser walk last year, which was the symbol chosen by Davide through his school homework.

We regularly speak about Davide’s school and life experience with Propionic Acidemia. Recently the New Aula Magna of his school was dedicated to Davide with a public ceremony. They also organize sport and cultural events including Golf tournaments, Cross country sky tournaments, Walking “through Davide’s paths” and a Concert of mountain chorus. They also received the status of “tax deductible donations association” to receive deductible contributions from people and companies. Paola has begun to print ceremony cards for marriages, Holy Communion, etc.

An update regarding research -- we signed an agreement with the Rome, Italy hospital, Bambin Gesu and on June 16th we’ll have interviews with the doctors to hire a researcher for 3 yrs with a total budget of 75000 euros (about $100,000 depending on the conversion rate).

Our objective is to continue in the following years with other researchers.

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Enamel Defects and Salivary Methylmalonate in Methylmalonic Acidemia:

Can the Tooth Fairy Help?

CW Bassim, National Human Genome Research Institute, National Institutes of Health, Bethesda, MD; J Sloan, NHGRI (National Human Genome Research Institute), National Institutes of Health, Bethesda, MD; CP Venditti, NHGRI (National Human Genome Research Institute), National Institutes of Health, Bethesda, MD; and TC Hart, National Human Genome Research Institute, National Institutes of Health, Bethesda, MD.

Our research team at the NIH has initiated studies on the dental health of patients with methylmalonic acidemia (MMA) and cobalamin disorders and recently published the first report describing these findings. (See reference note below.) We are very grateful to all the children and families that helped make these studies possible and believe we are now ready to further explore a new avenue of research that will have relevance to MMA as well as other organic acidemias.

Earlier this year, we described two issues that involve individuals with MMA: 1) enamel defects, and 2) saliva changes that may be associated with the condition. Problems with enamel development can occur when the body experiences a sickness during the time of tooth formation. Once developed, enamel remains fairly inactive, biologically non-responsive to internal stimuli, and functions without further remodeling. The presence of enamel defects, therefore, may be highly dependent on conditions occurring during the critical time of enamel formation and mineralization, such as mineral imbalances due to kidney problems. Enamel defects would then be a permanent indication of problems during this formation period. In a way, enamel development is like the growth rings on a tree. Also, since methylmalonate levels are extremely high in the bodily fluids (e.g. blood, urine) of individuals with MMA, we have examined methylmalonate levels in saliva. If saliva levels are correlated with blood and urine levels, saliva might also be considered in the future as another body fluid in which to measure metabolite levels. Saliva also helps to maintain a complex and balanced oral environment that is necessary for healthy gums, teeth, and possibly gastro-intestinal health. If high MMA levels are present in the saliva of individuals with MMA, this intraoral environment may be changed, altering the bacteria in the mouth and possibly affecting oral health. However, our impression is that these changes in enamel and saliva do not harm teeth or oral health, and people with MMA do not have higher levels of problems with their oral health than the greater population.

Enamel

Enamel is the white, smooth, outermost layer on teeth. It is the most mineralized tissue in the body and is made up of small calcium phosphate crystals oriented in a specific pattern to form enamel prisms. Enamel formation is a complex and highly regulated process involving the secretion and processing of proteins outside the cells called the extracellular matrix and the regulation of ion flow and crystal growth. Initial calcification of a primary baby tooth begins at 14-17 weeks in utero and enamel formation is completed at 1-9 months after birth. Permanent, adult anterior teeth begin calcification at 3-12 months and complete enamel formation at 4-7 years of age.

Enamel defects have been associated with many pediatric systemic illnesses during these critical periods of enamel formation. Enamel differences have also been described in patients with specific metabolic disorders, such as phenylketonuria, an amino acid disorder, and in those with mucopolysaccharidoses, a group of lysosomal storage diseases. However, the dental findings of individuals with MMA have not been thoroughly examined.

Enamel Defects

Enamel defects can be quite variable, and can appear as changes in the color, shape, thickness or smoothness of the tooth. They may appear as a discolored line, often bright white but sometimes yellow, brownish, or grayish, or as an area where the enamel is missing, as in a pit on the surface or a chip off of the surface of a tooth. Enamel defects seem to occur on the surfaces of the adult teeth of people with MMA to a greater extent than the general population. This is particularly true of individuals with mut MMA, where over half of those we evaluated had at least one tooth with an enamel defect. Individuals with mut MMA who also had higher blood levels of methylmalonate had more extreme enamel defects than those patients with mut MMA with lower MMA levels. Since individuals with mut MMA often have kidney problems, this may indicate that worse kidney problems during tooth development have led to the worse enamel defects. The baby teeth of the children with MMA appeared normal. The picture of the teeth of
Saliva

Saliva has the potential to be a non-invasive way to monitor an individual’s health or medical condition. Saliva is a complex fluid composed mostly of the product of the salivary glands in the mouth, with some contribution from fluid leakage around the gums of the teeth. It has important functions in digestion, in speech and swallowing, and in controlling disease-causing bacteria such as those that cause dental cavities.

Saliva MMA

Saliva from individuals with MMA has higher levels of methylmalonate than saliva taken from individuals without MMA. This seems to mirror the methylmalonate levels found in the blood of people with MMA: patients with high blood levels have correspondingly high salivary levels of methylmalonate. It is possible that this high level of methylmalonate would change the number and kind of bacteria living in the mouth. Interestingly, most of the patients we have seen with MMA have a limited amount of cavities or dental problems, which may partially be explained by the different salivary environment. More studies are needed to further understand these observations.

Future Studies

We are continuing to evaluate the teeth and saliva of individuals with MMA as they come to the NIH dental clinic and are thankful to all the patients who have helped with the research. Those who have not participated or visited the dental clinic during a trip to the NIH as part of the MMA and cobalamin natural history study will want to know that there is no drilling or painful prodding associated with the research dental assessment. We do try to collect saliva, photograph and X-ray the teeth, and look carefully at the oral cavity but it does not hurt! We hope to further characterize tooth and salivary changes in MMA and related conditions to improve our understanding of the dental consequences of MMA and to provide optimal dental care.

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May 7, 2009

Kathy Stagni
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Re: OAA grant to Dr. Charles Venditti of NHGRI

Dear Ms. Stagni,

I'd like to personally thank the Organic Acidemia Association (OAA) for its recent award of a $100,000 research grant to Dr. Charles Venditti, a tenure-track investigator with the National Human Genome Research Institute (NHGRI).

The use of the donated OAA funds will be governed by the terms of the existing Donor Agreement between OAA and NHGRI (execution date: April 14, 2004). Per the terms of that Agreement “…OAA donations will be used to support ongoing scientific research carried out by Dr. Charles Venditti…related to methylmalonic acidemia (MMA), a rare genetic and metabolic disorder” and the funds will be deposited into NHGRI conditional gift fund common accounting number (CAN) account 8363857 which is entitled “OAA-MMA Research Fund”. Questions about the Donor Agreement should be directed to Claire Driscoll, Director, NHGRI Technology Transfer Office (e-mail: edriscoll@mail.nih.gov/Tel. 301 594-2235).

The National Institutes of Health (NIH) is an agency of the United States (U.S.) Government organized for the conduct and funding of scientific research and is authorized to accept gifts under section 231 of the Public Health Service Act (42 U.S.C. 238). Authority to accept conditional gifts has been delegated to the Directors of the Institutes, Centers, and Divisions to support activities within their respective areas. The NHGRI is a Federal Agency of the U.S. Government and is recognized by the Internal Revenue Service as tax-exempt under 26 U.S.C. 501 (c)(3). Contributions to the NHGRI are tax deductible pursuant to 26 U.S.C. 170.

Thank you again for your generous support of NHGRI’s research mission.

Sincerely,

Alan E. Guttmacher, M.D.
Acting Director, NHGRI

cc: C. Venditti, K. Brown and P. Hylla
Cheyenne was born on August 31, 2005. She is now 3 1/2 years old and growing every day! Her story is like so many others with Glutaric Acidemia Type I that I have read through OAA. She started off healthy, but we had very limited newborn screening in Alabama at the time of her birth. She spent much of the first 2 years of her life in & out of the hospital for circumstances ranging from minor colds to severe seizures and vitamin/mineral deficiencies. She has undergone 4 surgeries and is expected to have another one soon. Her first was at 5 weeks for a g-tube. She has since had 2 depo-port and a Broviac. Her depo-port has stopped giving a blood return (although we can still use it to give fluids & meds while in the hospital) so we are looking at getting that replaced soon.

Cheyenne’s medications include Carnitine, Lamictal (for seizures), Panthothenic Acid, Riboflavin, CoEnzyme Q10, an Iron supplement and Flintstones multivitamins. Her diet consists of 14 grams of Protein per day, 99 grams of ProPhree and 15 grams of Glutarex-2. She is exactly 3 feet tall and weighs 34 pounds. She has short blonde hair and the bluest eyes you have ever seen! She has a crooked little smile and an infectious giggle.

Cheyenne started talking around 10 months (simple words like mama, dada, dog), but a seizure at 13 months caused her to digress. She now only says about 5 understandable words (mama, dada, bubba for brother, bye bye, night night). She attends the Chilton County Preschool Program—P.A.L.S. She is in an inclusion classroom and goes 2 days a week. We combine this schooling with 2 hours per week of speech therapy at the University of Montevallo. Although she is non-verbal, she is a very bright girl! She started “school” just after she turned 3 years old. At that time, they tested her verbal & non-verbal capabilities. She, as we suspected, scored in the 6-9month old range for verbal abilities; but her non-verbal score put her at the level of a 4 year old! She continues to learn more & more everyday. She’s hungry for knowledge. She knows all of her basic colors, how to pick out what she wants for dinner, how to show you how old she is, how to get dressed by herself and, to the doctor’s surprise, has been completely potty trained since just before she turned 3. I am so very proud of her!

Overall, Cheyenne is doing very well. I’m glad I haven’t had much to report! She went an entire year without a hospital visit due to her GA-1 (she did break her leg in October climbing on the lawnmower). Her doctors are very impressed with her accomplishments and unbelievable feats. She has beaten the odds and continued to grow, mature & develop into a vivacious, confident, precious, smart, beautiful 3 year old little girl whose hero is her big brother, Cody.

Cay Welch Honored with Distinguished Service Award

by Cay Welch

The weather was picture perfect in Pittsburgh PA Sept 17, 2009. It was my distinct pleasure to receive the Maxwell Schleifer Distinguished Service Award.

The award was given to me by Mark Stallsmith of Mass Mutual Insurance Company and Joe Valenzano of Exceptional Parent Global communications. So it was right after the singing of the national anthem on home plate of PNC Ballpark the pomp and ceremony began.

Mass Mutual is committed to recognizing Disabilities Awareness Night at the ballpark thru out United States. Pittsburgh is just one of the many stops on route across America for Mass Mutual and Exceptional Parent. There was a huge attendance of folks in wheelchairs that night. Included in that group was my son Michael Metil. I was able to accept this award along side my son who is wheelchair bound affected with Glutaric Acidemia, Type 1.

We all especially liked the fireworks display with the city of Pittsburgh as its backdrop. Somehow I just know Max would have loved it too. His devotion to children with special needs and passion is a gift I pray to continue to share for many years to come.
Kristin Rachel Propionic Acidemia, Age 9

Kristin, has it really been almost ten years since you came into our family? Our lives were so different a decade ago. Things were simple, understandable, and fairly predictable. Ten years ago I was pregnant with you in the middle of a hot Texas summer and running after your energetic two year old brother Eric. Eric had just potty trained....now he was a big kid, so of course that’s when he decided he also no longer wanted to take naps! Everyone was talking about stocking supplies for Y2K. By July I was already looking forward to late September when I would finally hold you and it would be cooler. One afternoon Eric listened to my tummy and told me you wanted to be named Tia, and then he set a small table with his bears and puppies for a tea party. After arranging the food he left one chair open and said it was for you, so everything would be ready when you came. On September 28, 1999 you were born, and although we were already aboard that ship to Holland we didn’t have a clue. Your first five months of life was a blur of confusion, worry and constant throwing up. Somehow you managed to grow even though you lost a large amount of your food each day.

Fast forward to February 24, 2000. After 5 months of sleepiness, lethargy and projectile vomiting this was the day you went into an actual metabolic crisis, but we had no idea what was happening to you. It was Eric’s birthday and he was turning three. We tried to wake you for the party, but you refused to wake up. It was really scary seeing you lay there so limp and unresponsive. We rushed you to Children’s Hospital in Austin, and I remember the ER physicians looking very concerned at you, then us, then you again. You were way too dehydrated for the amount you had thrown up. You were way too sleepy. You were very constipated. You had convinced each pediatrician since you were born that you just had baby reflux but inside I knew all along something was not right. No one listened. I had been nursing you, but decided to wean you to Similac and now you were really sick. After 3 days in the hospital the doctors said you had a stomach bug, but your dad and I knew that was highly unlikely. No one else in our family had been sick and you were cared for at home. So you were discharged and we went back home to feed you more high protein Similac.

Over the next 9 days you went downhill again, becoming sleepier and constipated just like before. You were switched to a lactose-free formula, then to soy. The soy helped, but by day 10 you were starving blankly into space and moving your head back and forth repeatedly. When I held you, you could barely hang on to my shoulder or lift your head. Your pediatrician called the hospital for the results of the urine organic acid test the ER physician had ordered. She told me to sit down.....you had propionic acidemia and most likely were going to be mentally and developmentally delayed. We had arrived in Holland.

The next few months were packed with doctor visits, driving to the metabolic clinic in San Antonio each month, learning about PA, and coordinating physical, occupational, speech and cognitive therapy visits. We learned quickly that insurance companies will do anything to avoid paying for medical formulas like Propimex and Prophree, even when state law mandates they do so. We learned how to calculate your protein, water and calories each day, and how to make your low-protein Propimex formula, which smelled and tasted horrible. I was amazed that you drank it from a bottle, but I guess adding carrots helped! I read everything I could about propionic acidemia. It didn’t sound good. The average lifespan of a PA child in 1999 was 5 years. At one point I decided not to read anymore journal articles so I wouldn’t hold a preconceived negative picture in my head of your future.

When you were 8 mos. old you caught your first cold and suddenly refused to drink your formula. We had been sitting up with you nightly in a rocking chair feeding you every 4 hours, but the doctors asked us to consider placing a g-tube, which we did. Your g-tube was changed to a g-button about 8 weeks later, and that button has saved us from many potential trips to the ER because we were able to give you calories and fluids at home. You still had major problems with refluxing, however, so after a 10 day stay in the hospital for RSV and more urging from the doctors we agreed for you to have a Nissen fundoplication when you were 17 months old. Your Nissen has worked so well, we have never regretted doing the surgery, but since you can’t burp or throw up anymore we do need to vent you before every feeding. We call you our baby whale and you laugh when you blow air out of your tummy!

Kristin, I won’t sugar-coat the next seven years. Life with PA has been hard. You threw up or wretched and gagged so many times in the early days that today when I hear any child gag my heart speeds up and I still get that panicky feeling of “Oh God, is this going to be a hospital run?”. Every time I heard you gag through the baby monitor during the nights I felt like Pavlov’s dogs running to your room to make sure you were not suffocating. Your dad and I were always second guessing our decisions, wondering if we were doing everything right. What if we goofed? What if we misjudged what you needed or misread your behavior? The early days were fast, frightening and incredibly stressful. I hardly remember Eric’s fourth year of life. The lack of sleep was a killer. It was hard not having my parents around since they both died years before you were born, and it’s been hard over the years seeing how many people shy away from you because you are different. If they would only get down on your level, take the time to connect to you, they would see what a sweet and talented child you are. You have developed into a patient playful little girl who often shows her impish sense of humor!

The biggest curve ball you threw us was your need for so much more water than the other PA’s we knew. From the start you required massive quantities of extra water or juice to stay hydrated and not develop ketones. We bolus fed you about every 30-45 minutes all day, every day, and you were on a pump overnight to get all your food and water into you. When you gagged we had to take out all stomach contents, get rid of the mucus and refeed you. We were so exhausted from your schedule we actually started the evaluation process for a liver transplant. Why were you so different from the other PA’s? We got our answer when you were 4 1/2 years old and were hospitalized for a stomach virus that Eric picked up at school. You barely stayed hydrated using a central line, and the attending physician told us you probably had diabetes insipidus to need those amounts of water. You were given DDAVP, an anti-diuretic hormone that helps the kidneys keep water inside your body instead of you pouting it all out so quickly.

Once you started taking DDAVP our lives have been so much better. You started initiating more activities since you felt better, and we have had far fewer gagging episodes. Instead of about 16 bolus feeds a day we are down to 8 or 9. It’s been easier to keep your ketones in check, and it’s been easier to keep home health nurses for care for you since you are not gagging all the time. Although you are very heavy, at least you have been stable over the last 4 years. Reducing your calories has failed numerous times, but we are trying a new approach by very slowly reducing fat calories to hopefully keep you from gaining more weight. The cocktail of Coenzyme Q10, DHA, vitamin E, B-vitamins, carnitine and sodium succinate have helped your mental growth tremendously. You were hospitalized last summer because you had 3 seizures which was caused by not enough sodium in your diet. The DDAVP, salt and water concentrations are another factor we have to keep tightly regulated in addition to your PA.
Update:

Sam

Isovaleric Acidemia, Age 15

(Original story in the Winter, 2005 OAA Newsletter)

My pregnancy and delivery with Sam were normal. After he was born in 1993, he started to vomit, and it became progressively more over his first few weeks and was projectile. He had some outpatient tests for GI disorders, and I was advised to try several different formulas. Nothing worked, and at 4 weeks of age he was still at his birth weight, and he was admitted to the hospital, diagnosed with a urinary tract infection.

He was treated for the urinary tract infection, but the doctors quickly realized that there was something else underlying wrong, as he was not getting better, and was termed as “failure to thrive”. He was tested for everything imaginable, and finally after 3 weeks in the hospital, he was diagnosed with IVA. He did not have the classic symptoms, never went into a coma, and did not have the “sweaty feet” smell.

He immediately went on a combination of Prophree formula and Similac, along with glycine and carnitine, and started to grow. He did very well, right away. He only had 2 incidents in the hospital after that time-both flu related from vomiting and dehydration, once at about a year, and the other time about 2.5 years, with one night in the hospital. Both times, after going on an IV, he quickly turned around, and was able to keep fluids down.

At the age of one, he started on I-Valex formula, which he remained on until about 5 years old. We then switched to Xleu Maxamaid. He wasn’t having any problems on the I-Valex, but I couldn’t get him off of the bottle. He knew he was too old for a bottle, so wouldn’t drink it in front of anyone but family, but that still the only way he would drink his formula. The Xleu is more like a juice (it’s compared to Tang, whereas I-Valex looks more like milk), and with the switch, I told him we were throwing out his bottles, and luckily it worked. We mix it with Gatorade, and his compliance is good with it, which I hope continues as he gets older.

He has serious food issues. There are many foods he can have that he won’t touch. He only eats carbs, and is extremely particular with all aspects of food, including preparation, look, etc. He is very hesitant to try anything new. He won’t touch any fruits or vegetables, and lives primarily on pizza (cheese removed), pasta, French fries, cereal, crackers, bread, mac and cheese (light cheese), and chips. I hope he will expand his diet as he gets older, but he truly has some major food aversions. I worry health wise about his unbalanced diet, and also socially, because all social functions revolve around eating, and I don’t want him to feel that he doesn’t want to participate in things because he doesn’t eat much. As a kid, it really doesn’t affect him, because all kids like pizza, and French fries! He sees me eating a lot of salad and fruit, but unfortunately it doesn’t interest him!

He is currently in the 9th grade, does very well in school, and has a quick wit. He is a good athlete, with a passion for baseball.

Kristin, you have come such a long way from that pale, limp 5 month old baby who went into a major crisis because Texas did not screen newborns for PA in 1999. You have beaten the odds, and in a few months you’ll be ten. Although you are globally delayed you are a very happy child! You have learned how to crawl, knee-walk and climb into your bed, chairs and sofas. You carry toys from one room to the next, you roll balls and push cars, which is especially fun with Eric, and you can now sit in the bathtub all by yourself without being strapped in a bathchair. Just recently you learned how to open and close the foot rest of the Lazy-boy in the gameroom with either the side handle or your toes! OK, you earned it. The chair is officially yours to watch your favorite CD’s like Zoboomafoo, Clifford and Bear in the Big Blue House. You have learned how to point to which CD you want to watch, and you love your interactive push button toys and books. The more music the better, and this past year you started singing parts of songs, on key to boot! You like us to sing the songs back to you, help you play them on your toy piano, and you are vocalizing more and more sounds. You have several hand gestures to show us: “down, go outside, NO!, enough, I’m mildly irritated, and I’m going to scream if you keep doing that!” You have distinctive sounds when you are either thirsty or hungry, and you have a belly laugh that makes everyone smile. You learned how to draw with a crayon this year, to open and close the refrigerator door, and you like to match the farm animals on your wooden puzzles. Your favorite thing is to have Daddy give you your bath and to go riding in the van over bumpy roads so we can all say “whee!” You simply love a good g-force around a curve! We are working towards identifying flash cards, alphabet letters and pictures in books, and you have several communication devices with recorded messages you can select. Your favorite places to visit are the beach and our family farm. You have gone to Padre Island in Corpus Christi every summer of your life, and you adore walking along the seawall with the breeze blowing in your hair. You love playing with your cousins Kasey, Chandria and Yesinia and will now give and take toys from them. You love your splashing pool outside on the deck, but I think your absolute favorite thing is to talk back and forth with Daddy and make funny faces so he’ll laugh.

One of the best developments for our family is that in spite of the ongoing stress and chaos, your brother has developed into a truly compassionate and talented young man who has excelled at school in every grade. This year Eric was voted Best Citizen and was chosen to be in the National Junior Honor Society in 6th grade at his middle school. To bring some meaning out of this PA experience I have felt a personal need to be involved with PA research activities. In order to help our kids and future kids I have spent quite a bit of time serving on the boards of both the Organic Acidemia Association (OAA), and the Propionic Acidemia Foundation (PAF) over the past 8 years. Both of these organizations exist to serve children with organic acidemias and to facilitate research into PA and other metabolic disorders. One day, Kristin, there will be a cure for PA. Hopefully you and I will have helped to facilitate research into PA and other metabolic disorders. One day, Kristin, there will be a cure for PA. Hopefully you and I will have helped somewhere on that path.

So Kristin, my beautiful little girl, we’ve all come a long way. Your needs have pushed us farther than we ever thought we could handle, and although we have had some scary moments we know that God provides in interesting ways, and in His timing. Let’s go play! We love you.

Mommy, Daddy and Eric

Lisaw

jsboecker@austin.rr.com

512-394-0977

Austin, TX 78739

10305 Hansa Cove

Janice and Russell Boecker

Mommy, Daddy and Eric
Cambrooke Foods – June 2009

Cambrooke Foods has been busy making your favorite products better. Try the new and improved Chocolate Cha Chas, the low protein mini chocolate bars that taste and melt like real milk chocolate. They’re back and better than ever!

The new Mini Pockets™ are filled calzone-style snack or meal items that are quick, heat-and-eat items. They’re great for busy families looking for home-made taste in a hurry. Two varieties are available: Pizza and PB&J.

Celebrate summer with delicious Brooklyn Dogs™, veggie hot dog taste-a-likes. These fat free, hi fiber, low protein dogs are sure to be a family favorite! They contain 32 mg of DHA Omega-3s per serving, too.

Ingredients for the Brooklyn Dogs: Yuca (Corn Flour, Palm Oil, Sugar, Water, Salt), Inulin, Water, Sweet Potatoes, Red Bell Peppers, Artificial Flavoring (Hydrolyzed Soy Protein, Sodium Chloride, Silicon Dioxide, Flavors, Partially Hydrogenated Soybean Oil, & Glycerol Triacetate), Caramel Color (preserved with sulfites), Salt, DHA Algal Oil (High Oleic Sunflower Oil, Sunflower Lecithin, Tocopherals and Ascorbyl Palmitate as Antioxidants), Natural Lamb Casing. (Contains soy)

The protein is 0.5 g per 2 oz.“dog.”

To order:
Toll-free 866 4 LOW PRO / 866 456 9776
www.cambrookefoods.com
orders@cambrookefoods.com

Mac-A-Weenie & Cheese

Ingredients

- 1 box (7 oz) CBF Pasta Duets™ - Mac & Cheese
- 1 cup non-dairy creamer
- 1 Tbs unsalted butter
- 2 CBF Brooklyn Dogs™

Directions

- Pan fry the two Brooklyn Dogs™ and cut into 1/2 inch slices.
- Prepare Mac & Cheese according to package directions.
- Blend the “dogs” and Mac & Cheese into a greased casserole.
- Bake at 350º F for 10-15 minutes or until golden brown.

Suggestion: Cover casserole with toasted low protein breadcrumb topping.
Serving size: 185g (Makes 3 servings)
Protein: 1.1g per serving
### Upcoming Events

<table>
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<tr>
<th>Date</th>
<th>Event</th>
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<tr>
<td>June 22-27, 2009</td>
<td>15th Annual Metabolic Camp</td>
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<td>Department of Human Genetics</td>
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<td>Division of Medical Genetics-Metabolic Nutrition Program</td>
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<td>Emory University Atlanta, GA</td>
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<td>June 24-29, 2009</td>
<td>United Mitochondrial Disease Foundation Symposium</td>
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<td>Mitochondrial Medicine 2009: Capitol Hill</td>
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<td>Scientific Meetings: June 24-27, 2009</td>
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<td>Family Meetings: June 26-27, 2009</td>
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<td>Special Day on the Hill: June 25, 2009</td>
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<td>July 10-12, 2009</td>
<td>Children’s Hospital of Boston Teen Challenge Camp</td>
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<td>Designed for Teens ages 13-18 with PKU or other metabolic disorders</td>
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<td><a href="http://www.childrenshospital.org/newenglandconsortium/teenchl08.pdf">http://www.childrenshospital.org/newenglandconsortium/teenchl08.pdf</a></td>
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<td>August 29 - September 2, 2009</td>
<td>11th Annual International Congress on Inborn Errors of Metabolism</td>
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<td>San Diego, CA</td>
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<td>The Organic Acidemia Association and FOD Family Support will</td>
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<td>be one of the many exhibitors present at this very important</td>
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<td>meeting. Deb Gould from FOD Support and Kathy Stagni from OAA will</td>
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<td>be representing our families in San Diego.</td>
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<td>September 4th-7th, 2009</td>
<td>6th Annual PKU Family Camp</td>
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<td>Washington Family Ranch</td>
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<td>Formerly Wildhorse Canyon</td>
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<td><a href="http://washingtonfamilyranch.younglife.org">http://washingtonfamilyranch.younglife.org</a></td>
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<td>Calling all Campers!!! The PKU Northwest Alliance invites</td>
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<td>individuals with PKU and other low-protein diets, plus their families</td>
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<td>and friends, for a long weekend of fun at the Washington Family</td>
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<td>Ranch in Central Oregon. Join us over Labor Day weekend (Friday-</td>
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<td>Monday) for a weekend your family will never forget! All ages are</td>
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<td>welcome.</td>
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<td>For general inquiries, contact Anne Bedney at 503-232-2305 or</td>
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<td></td>
<td>email <a href="mailto:pkunwa@gmail.com">pkunwa@gmail.com</a></td>
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<td>For registration questions, contact Stacie Hofman at 503-405-7710 or</td>
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<td>email <a href="mailto:shofmann@wfr.younglife.org">shofmann@wfr.younglife.org</a></td>
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<td>September 18th, 2009</td>
<td>IOGA (International Organization for Glutaric Acidemia): Golf Tournament</td>
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<td>IOGA will host the seventh golf benefit in Latrobe, Pa. Latrobe</td>
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<td>Country Club along with Mr. Arnold Palmer have been a partner</td>
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<td>to our non profit making it possible for us to do this fundraiser.</td>
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<td>This is IOGA’s ONLY fundraiser. What we are asking of all our</td>
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<td>audience and families and friends is to PARTICIPATE.</td>
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<td><strong>Hole Sponsorship:</strong> we need 18 to be exact they cost $150.00</td>
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<td><strong>Golfers:</strong> single or sets. We will gladly pair up people. There is</td>
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<td>no problem if you are by yourself. LAST YEAR WE HAD 17 4-SOMES.</td>
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<td>This year my goal is 25 4-somes.</td>
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<td>Should you want a brochure or letters of solicitation I have them</td>
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<td>just ask me, give me your address and I will mail them to you.</td>
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<td>Again we appreciate all the help. We welcome all of you to come</td>
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<td>to southwest Pennsylvania and join us on this date.</td>
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<td>Friday evening at 5pm our activates will begin.</td>
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<td>Located at: The clinic for Special Children in Strasburg, Pa. Meat</td>
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<td>and beverages will be provided. Simply bring along a dish to</td>
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<td>share with the other families. This is our opportunity to visit other</td>
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<td>GA families from near and far.</td>
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<td>Games and dinner are a part of our evening. There is no charge for</td>
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<td>this event. Its part of our mission of fellowship with one another.</td>
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<td>It does help to have a response. So please let us know if you will</td>
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<td>be attending so we can prepare enough food.</td>
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<td></td>
<td>Contact Cay Welch, for any of your questions 724-459-0179.</td>
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</table>
Dr. Charlie Roe recently retired from the Institute for Metabolic Disease …but he’s not ‘retiring yet’!

Thank you Dr. Roe for your dedication to the field of metabolism!

We will keep you posted on his ‘beyond retirement’ work once this information is received.