

Propionic Acidemia Foundation

VOLUME 1, ISSUE 6

SPRING 2009

PA FAMILY DAY 2009 JULY 25

Please join us for this year's annual PA Family Day. This year the Koehl family is opening their home to us as first time hosts for the event. All PA families and friends are welcome to attend!

- Lunch and tasting of low protein foods
- Update on PAF activities/board meeting
- Meet new people and visit with old friends
- Outdoor fun for the kids (including swimming!)

The Koehl Home
7670 McVile Road
Burlington, KY 40502
11 am – 5 pm

RSVP to Jill Franks at PAF by July 15th
Toll Free Phone: 1-877-720-2192
Email: paf@pafoundation.com
Contact PAF for more information.



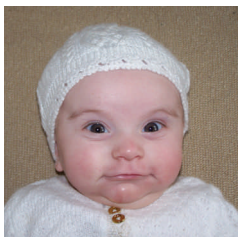
Julie, Hogan and Harper Koehl

For the first time PAF may have to turn down a research project due to increased financial needs by researchers and not enough funding available through PAF. We are so grateful for more interest in studying PA and hope that funding will become available to further search for new treatments and a cure.

INSIDE

FAMILY STORY: LOGAN	2
FAMILY STORY: ANGELICA	3
FAMILY STORY: ANSON	4
FEEDING TUBES: NG & G-TUBE	5
FUNDRAISING UPDATE	6
CARBAGLU STUDY UPDATE	7
DONATION FORM	7

FAMILY STORIES INSIDE THIS ISSUE:



ANGELICA



ANSON



LOGAN

MISSION: The Propionic Acidemia Foundation is dedicated to finding improved treatments and a cure for Propionic Acidemia by funding research and providing information and support to families and medical professionals.

VISION: To create a future where Propionic Acidemia can be prevented and any affected individual can be cured and live a productive life.

FAMILY SPOTLIGHT - LOGAN'S "MILD" PA

When my ex-husband called to tell me our son, Logan, was being transported by helicopter to Children's Hospital of Wisconsin in Milwaukee I was in disbelief. Logan was a normal, healthy, active 15 year old. He was a sophomore in high school, eager to get his driver's license and training for the school tennis team. Yes, he had been sick for a couple weeks with what seemed to be an upper respiratory infection and then possibly even asthma, but when I heard he had cardiomyopathy, I couldn't believe it. I knew all too well this was an enlarged heart.

Our oldest daughter, Justine, also a healthy child had been diagnosed with the same condition 20 years earlier when she was six years old. At that time the doctors told us they believed a virus attacked her heart. They could find no other cause and said this sometimes happens. There was nothing they could do for her. The subject of transplant was brought up, but quickly dismissed. Medical technology didn't make this a realistic option 20 years ago. Within seven months she died. We had no other children at the time, but we were assured this should not be a concern for any future children we may have.

Two years later Latrece was born, and then three years after that we had Logan. Both were healthy and routine wellness visits to their pediatrician never indicated there was any cause for concern.



Justine, age 6

This was not supposed to happen again! Why? Within the next couple weeks of April 2005 we were questioned by doctors trying to figure out what the connection was between Justine's and Logan's heart condition. We also found medical advancements had made treatment much different.

Within two days, Logan was listed for a heart transplant. But his condition was deteriorating quickly and he needed to have surgery to put him on a left ventricular assist device (LVAD) to support him while he waiting for transplant. Surgery went fine, but over the next few days we realized something was wrong. Logan was in status with subclinical seizures and



not waking up. He had no previous history of seizures. Now neurologists were consulted. They could not control his seizures with standard medication techniques and as an only option decided to try putting him in a deep drug induced coma. They started sending blood, muscle, skin and spinal fluid specimens for testing to laboratories throughout the country. Our daughter, Latrece, was also tested. She was fine. Some of the initial results pointed to the direction of some type of metabolic disorder, but we were told it would take months to know exactly what we were dealing with. In the mean time, while Logan was still in a coma, genetics was called in. They recommended starting Logan on a "cocktail" of supplements that would be beneficial to someone with his suspected range of disorders. Finally after a month, something worked. Logan was awake without seizures. Now he required

intense physical therapy to get his body strong enough to be re-activated for transplant. June 18, 2005 the day came. This surgery went remarkably well and a week later we received the lab results we had been waiting for: Propionic Acidemia (PA). Logan had an elevated C3 level in an acylcarnitine profile, and his urine tests showed chemicals that suggested it was PA. I had no idea what this was, but spent several weeks searching for information. Logan just didn't seem to fit. He had never been sick as a baby or child like these other unfortunate children I was learning about. I was told Logan has a mild (late onset) case. I felt I should be thankful, but was rather puzzled by what a "mild" case was capable of. Skin and heart muscle were sent away for enzyme analysis for propionyl CoA carboxylase that showed he has 4% enzyme activity. While his enzyme activity is low, most children that are sick as newborns have 0% activity, which is likely why he is "mild" and didn't have any symptoms until his cardiomyopathy appeared as a teenager.

Logan left the hospital a month after his transplant and with the help of tutors was able to catch up on his studies and complete high school with his class. In many ways Logan has been a normal teenager. He does have many doctor appointments with cardiology, neurology and genetics and takes many medications and supplements, but *(continued on bottom of page 3)*

FAMILY SPOTLIGHT - ANGELICA



Our daughter Angelica had her first 'metabolic crisis' at three days old. Within twenty-four hours of bringing her home from the hospital after birth, she had become so

lethargic and unresponsive that we had no choice but to contact the nurse on call, who instructed us to immediately call an ambulance. The staff in the emergency room (thankfully, we live less than ten minutes away from the fantastic resources of the University of Michigan Hospital) were able to stabilize her to a degree, but were very troubled by her ammonia levels and other test results, though they did not know the cause at that time. A conclusive diagnosis of PA was reached by the following day, and after another harrowing four days in intensive care undergoing hemodialysis she was moved to a moderate care wing. We were able to return home about a week after that, once she had regained enough strength, and more importantly, her appetite.

Now she is a little over four months old, and while there have certainly been bumps in the road, it does not seem

like we could have asked for a much better situation. She has an incredibly hearty appetite for her special formula ("power milk") and is the sweetest, happiest, most easy going kid you could ever hope to see, and has even been letting her mom and dad sleep through the night, pretty much since we came home! She is tough (and stubborn) as well, having survived intensive dialysis at such a young age and showing no signs of damage from the ordeal.

The pediatric genetics staff at the UM Mott Children's Hospital are a great resource, and having them so close at hand is an incredible luxury that we definitely don't take for granted. We are also blessed with a very competent and understanding daycare provider-- they take everything in stride, whether it is diluting formula as needed, providing medications, or even ketone testing. With everything we have at our disposal, the inevitable problems we are going to encounter do not seem nearly as daunting as they once did.

Best Regards,
Joe Stageman and Toula Saratsis
(and Angelica)



"LOGAN", CONTINUED FROM PAGE 2

he had been doing well. He has been attending Tech school and shared an apartment with several roommates.

Unfortunately, on January 30, 2009 Logan was admitted to the hospital again. He is now dealing with severe rejection and is in need of another heart transplant and possible kidney transplant due to poor perfusion. He again is being supported by a ventricular assist device (BIVAD), dialysis and a tracheotomy. He is not healthy and strong enough to be listed for re-transplant yet, but we are hoping he will be soon. Genetics continues to be consulted to determine how his metabolic disorder plays a role in his symptoms and treatments.

In the last several years our family's DNA mutation have been tested. It was confirmed that Logan's father and I are both carriers of PA, and our daughter Latreace is also a carrier. I am sure there are other cases like Logan's and

I often wonder if Logan's condition could have been prevented if he was diagnosed and treated early in life. Newborn screening for propionic acidemia is now done in most states in the US, but it is unknown whether infants with mild PA will be detected and treated to prevent sudden cardiomyopathy.

Special thanks to Amy White, MS, CGC for assistance in preparing Logan's story, and thanks to the entire talented and dedicated staff at Children's Hospital of Wisconsin, Milwaukee.

Debra Laack, mom
Logan (19) PA

To follow Logan's story:
caringbridge.org/la/logan



FAMILY SPOTLIGHT - ANSON

Anson was diagnosed with PA when he was 2.5. His mother noticed once she stopped breastfeeding and put Anson on whole milk that his health seems to slowly get worse. He would sleep more and it became harder and harder for him to have bowel movements. When his growth started to become stunted and he would scream while having bowel movements, his mother brought him to his doctor and had him tested. At that time, his family doctor happened to remember reading about organic acidemia's in a paragraph in his medical book and had Anson tested. Once he was diagnosed, Anson's mom was put in touch with Dr. Packman from UCSF.

After being in and out of the hospital for several years and testing many medications, Anson finally seemed to stabilize and his family was able to settle down a bit. At around age 12, Anson felt like he didn't fit in and that to make it better, he started to ignore the fact that he had PA and thought it would go away since he had been out of the hospital for many years. He stopped taking all medication, either hiding it from his mother, or refusing to take it altogether. Anson attended jr high and part of high school with no complications. He ate what he wanted and tried to forget his PA. He made the track and soccer team and had become very athletic.

A couple of days before Anson's 18th birthday, he was sitting with his mom and step-dad (mom was an EMT and step dad is a volunteer firefighter) when his heart



Anson with his two children

went into b-fib and he stopped breathing. His mother and step-father kept him breathing until he was life-lighted to a hospital where they put him in a chemically induced coma to figure out the problem and how to fix it. Doctors told Anson that the amino acid in his system had built up as fatty tissue around the heart causing the b-fib. It was also discovered that Anson had arrhythmia and an enlarged heart. A pacemaker/difribulator was put in and he was put back on his medications.

About a year and a half later is when we met. We dated 6 months and were married. One our one year wedding anniversary, we found out we were pregnant with our 1st child. She is now 4 and we have a 21 month old son. They are both unaffected by PA. (neg. in newborn screening) Anson now takes his carnitor in addition to a betablocker. He is currently able to eat a little under the normal RDA of protein for an average sized man. His blood still shows signs of having PA present, but currently all of his levels are normal, and his heart is now at normal size. He works as a travel nurse recruiter locally and we have a marketing business on the side. Anson loves life and his family and is committed to showing God's goodness no matter what the circumstances.



Anson and Colleen with their children

—Colleen Juul

Propionic Acidemia related Heart Conditions include:

Prolonged QT is an abnormality in the heart's electrical system. The QT interval is the section at the end of a heart beat during which time the heart is a rest and preparing for the next beat. If the QT interval is lengthened, there is a risk for the next heart beat to start before the heart is actually ready to normally accept the activity. A severe abnormal heart rhythm can then develop causing dizziness, shortness of breath, sudden loss of consciousness and may cause sudden death.

Cardiomyopathy is a serious disease in which the heart muscle doesn't work as well as it should. The heart muscle is weak and doesn't pump effectively. Patients are at risk for heart failure, abnormal heart rhythms, and death. Other complications may include blood clots, high blood pressure, difficulty breathing and stroke.

It is strongly suggested that children with PA have at least a yearly visit with a cardiologist. These cardiac conditions often go undiagnosed until a problem occurs which could be too late. There are treatments and medications to help try to prevent some of the complications of these conditions, but there is not a cure.



—Julie Harding, RN

FEEDING TUBES: NGs AND G-TUBES



Gwen with a nasogastric (NG) tube

longer time such as a year or more; then a gastrostomy tube (G-Tube) would be the best choice.

The NG tube is easy to insert and can be done in the office or even at home. Though a skilled nurse often places these tubes, even parents can be trained to do this. The tube itself is a long flexible tube made from a soft plastic (not latex) with a feeding port at one end and a side hole at the other end which empties into the stomach. The NG tube is introduced into a nostril and pushed down to the stomach. If the child can cooperate they may be asked to swallow water as it is done as this helps it move down the esophagus into the stomach. It is removed simply by pulling it out. The drawbacks with long term use are possible sinus problems, discomfort (most children are OK with NGs in the short term) and interference with eating in some children.

The G-Tube on the other hand is a minimally invasive procedure. It can be placed surgically or more often endoscopically. This latter procedure is very simple and takes less than 15 minutes. Done under anesthesia, an endoscope (a long and slender tube which lets the physician see into the stomach) is pushed through the mouth into the stomach. The spot for the G-Tube is then chosen. At this time, after cleaning the abdominal wall on the outside; another physician inserts a large needle into the stomach.

Some children may be unable to eat enough food orally. When this happens either a temporary or more permanent feeding tube may be needed. A nasogastric (NG) tube is typically used for a temporary period which could be from a few days to several months. If feedings are needed for a

A suture line is threaded through this needle and the end is grabbed by the endoscope inside the stomach and the endoscope and suture line are pulled out of the stomach and mouth. At this end the G-Tube is attached and from the abdomen the suture line is pulled, drawing the G-Tube into the mouth and down to the stomach. The skin on the abdomen is nicked with a scalpel and the G-Tube is pulled out from the stomach through the nicked opening. The back end of the G-Tube is mushroom shaped and is now up against the stomach wall. Feeding ports are attached and the G-Tube is ready. Once the opening is well healed (about 2 months) then the G-Tube can be replaced with a low profile "button" G-Tube which is flush against the skin. This tube is usually replaced every 3-6 months depending upon the tube type and can be done in the office or even in the home.

The decision for a feeding tube is a serious one. Used appropriately, feeding tubes can provide nutrition that the child is not getting. Specialized formulas that a child may not take orally can be given and if the child is having trouble tolerating larger volumes of food they can be fed over longer periods of time with smaller amounts, even while the child is engaged in other activity.

Dr. R. Charles Dumont
Associate Professor Pediatrics
Program Director, Pediatric Metabolic Support Team
Loyola University Medical Center, Maywood, IL



Talli with a gastrostomy tube (G-Tube)

Publication Note: The PAF Newsletter is published twice a year. Readers may subscribe by writing to PAF, registering online or calling 877-720-2192. Letters and article submissions are welcome for consideration and may be sent to paf@pafoundation.com or mailed to Propionic Acidemia Foundation 1963 McCraren, Highland Park, IL 60035. If you would like to be removed from our mailing list or receive the newsletter via email, please contact us. We are deeply appreciative to Publishers Printing Company for donating the printing costs of the newsletter.

PAF FUNDRAISING SPOTLIGHT

Current Events and Campaigns

May 30 Poker Run for PAF, Columbus, Ohio Starts at Groucho's Lounge, 5770 Columbus Square between 1:00-2:30 p.m. Contact Michelle Ellis at Michelle@AllisonForACure.com for more information.

June 27 Corn Hole Tournament & Hog Roast, Pomeroy, Ohio
Contact Julie Howard at jbhoward_99@yahoo.com for more information.

July 5 Climbing for a Cure, Seattle, Washington Michelle Burns is doing a Mt. Rainier climb in Seattle, WA, in honor of her daughter Kaitlin. Donate directly to PAF online, by mail or through <http://www.firstgiving.com/climbingrainierforacure>.

September 19 Tailgate Party & Corn Hole Tournament, Columbus, Ohio
Email Jen Mouat at Jen@GwenForACure.com for more information. (Rain date Oct. 3)

Past Events and Campaigns

Quicken Loans Jeans Day— raised \$1283.50, Stageman-Saratsis Family (see pg. 3)
Every so often our office holds a Friday 'jeans day'. Our sales force is allowed to wear jeans for a small (typically \$5) donation to various causes. When I sent a picture of Angelica to the gal who organizes those events at work, it took her all of five minutes to reply to my email and let me know that she was going to reserve a date. That's the kind of effect our little girl has on people. She is actually as sweet as she looks.

Marissa's NYC Marathon— raised \$9366 for PAF and \$565 for OAA, Marissa Cotrina
I had to wake up at 5:00 a.m. to be at the Ferry at 6:00. Everything closed at 7:30 am and had to wait with all the runners for 3.5 hours until my start time. Really, really cold, no tents, no nothing. Luckily, I had been told to be prepared for the cold (43 degrees) and wore 3 layers of disposable clothes (heavy smell of basement there!!!). Everybody wears clothes that you then leave at the start. Volunteers collect them all and give them to charity. Never thought of that, eh? People carried even sleeping bags, blankets or whatever to feel warm. The race: FANTASTIC!!! What an experience.

I had the greatest time. The public was awesome, everybody cheering and feeling for you. Children line along the race course to give you high-five, bananas, tissue, candy... whatever they thought might help. I had so much fun!!! Statistics: TIME: 4:26:01 (all in one piece!) PLACE: 20932 out of 38356 By gender: 5274 out of 13002 women By age: 997 out of 7069 Fundraising: We collected \$9,366 for Propionic Acidemia Foundation and \$565 for Organic Acidemia Association THANK YOU all for your inspiration. Thanks also to all the families that contributed. Really, it meant a lot to us. We were thrilled on how everything work out and it was an amazing experience. I am sending a couple of pictures pre-race for you to see my look (really, not a fashion show here!) and my "special" shirt for the occasion!

Marisa, mom to Gabriel, PA 6 (who watched mommy "run so fast" on Sunday!) NYC, New York

Scrapbook Day for PAF— raised \$1700, Michelle Ellis, Columbus, Ohio
Michelle hosted the scrapbook day at Archiver's. Participants paid for workshop time (all to PAF) and enjoyed door prizes, food and great friends up to 12 hours!

Memorial Donations for Elliott Kirby— raised \$605, in honor of great-grandson, Hogan Koehl.

GoodSearch
Raised \$554

iGive.com
Change online shopping for good.
Raised \$1873

MissionFish
Raised \$705

eScrip
Raised \$129



N-CARBAMYLGLUTAMATE STUDY UPDATE

The N-Carbamylglutamate study is carried out at Children’s National Medical Center in Washington DC and Children’s Hospital of Philadelphia, by a team led by Drs. Mendel Tuchman and Marc Yudkoff. The purpose of this study is to investigate whether N-Carbamylglutamate (NCG, Carbaglu) can improve urea cycle function and decrease blood ammonia in Individuals with propionic acidemia, methylmalonic acidemia as well as urea cycle disorders such as NAGS deficiency and CPSI deficiency

Elevated blood ammonia associated with several rare inherited disorders frequently causes mental retardation, developmental disabilities and death.

carbamylglutamate. Participants with propionic or methylmalonic acidemia showed improvement of ammonia, urea cycle function, and amino acids, but to a lesser degree than patients with NAGS deficiency.

In 2008, additional funding was awarded by the National Institutes of Health to support this project. Plans for 2009 include enrolling further participants and starting an NCG trial for hyperammonemia and

hyperinsulinism syndrome at the Children’s Hospital of Philadelphia. Also, although NCG is not yet available for clinical use in the United States, an application to the Food and Drug Administration will be submitted this year by Orphan Europe, with the hope that this medication will be made available for clinical use soon.

Nicholas Ah Mew, MD FRCP(C)
 Biochemical Genetics Fellow
 Children’s National Medical Center



Grace-Marie Housley in D.C. following her participation in the study.

Since the beginning of the study, 7 patients have been enrolled: 4 with propionic acidemia, 1 with methylmalonic acidemia and 2 with NAGS deficiency.

Participants with NAGS deficiency showed normalization of urea cycle function and ammonia after treatment with N-

For more information on clinical trials visit www.clinicaltrials.gov and search "Organic Acid Disorders" or "Propionic". You may also contact PAF at 1-877-720-2192 or paf@pafoundation.com

Help Us Find The Cure!

Name _____
 Address _____
 City, State, Zip _____
 Phone _____
 E-mail _____

Please send an acknowledgement to:
 Name _____
 Address _____
 City, State, Zip _____

Enclosed is my contribution of \$ _____ in honor of/ in memory of _____

If you work for a company that has a matching program, please include the matching form.

Please mail your check made payable to: Propionic Acidemia Foundation 1963 McCraren, Highland Park, IL 60035

Thank you for making a difference.

SEARCHING FOR A CURE
HOPE FOR OUR CHILDREN

Propionic Acidemia Foundation
1963 McCraren Rd.
Highland Park, IL. 60035



Phone: 1-877-720-2192 toll free
Fax: 1-877-720-2192
E-mail: paf@pafoundation.com
Website: www.pafoundation.com

PROPIONIC ACIDEMIA FOUNDATION

Board of Directors

Jill Chertow Franks, President
Brittany Smith, Secretary
Janice S. Boecker
Jennifer Mouat
Michael Rosenson

Medical Advisory Board

Gerard T. Berry, M.D.
Barbra Burton, M.D.
Pinar T. Ozand, M.D., PhD
William Nyhan, M.D., PhD
Mendel Tuchman, M.D.

Thank you to our corporate sponsors:



GeneDx

Mead Johnson



Innovation in Nutrition

Thank you **Publishers Printing Company** for contributing the quality printing of the PAF Newsletters.

We would also like to extend our warm thanks to Michelle Burns for her years of service on the PAF board.

The Propionic Acidemia Foundation Newsletter is designed for educational purposes only and is not intended to serve as medical advice. The information provided should not be used for diagnosing or treating a health problem or disease. It is not a substitute for professional care. If you suspect you or your children may have Propionic Acidemia you should consult your health care provider.