

# Propionic Acidemia Foundation

VOLUME 1, ISSUE 10

SPRING 2011

## TGBK Basketball Shootout

Friends Brandon Keller and Tyler Gold, both seventh-graders at Caruso Middle School in Deerfield, IL, created the TGBK Basketball Shootout to raise money for three foundations with which each student had a personal connection. The three foundations they raised money for are The Pediatric Brain Tumor Foundation, The Propionic Acidemia Foundation, and The Jill Landau Triple Negative Breast Cancer Foundation.

The event raised \$6000 total and \$1500 for Propionic Acidemia Foundation.



Brandon & Tyler at the 2010 "Shootout"

TGBK Shootout was attended by 160 kids and included five skill areas: free throws, lay-ups, dribbling, 3 point shots and knock-out.

Debbie & Robert Keller and Nancy & Adam Gold (Brandon's and Tyler's parents) have embraced their efforts, "We're so proud of the boys, all of their hard work paid off."

Brandon and Tyler were also pleased with the results, "It was so much more than we thought it would be."

Congratulations on a successful event!

### In Memory of Lauren Delima



June 14, 2001-  
October 13, 2010

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**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.

## PA CONSENSUS TREATMENT CONFERENCE

Date: January 28th-30th, 2011

Location: Washington D.C.

On January 28-30, a group of 16 metabolic physicians, research scientists, metabolic dieticians, a transplant surgeon, parent group representatives, and neurologist met in Washington DC to hold the first Propionic Acidemia Consensus Treatment Conference. The meeting was coordinated and organized by Dr. Kimberly Chapman and sponsored by Children's National Medical Center. Using input and questions from clinicians and parent groups, the two-day session focused on statements about diagnosis, acute and neonatal management, natural history of the disease, chronic complications and health monitoring and future research.

The individuals attending included: Dr. Nicholas Ah Mew (Children's National, Washington D.C.), Dr. Anupam Chakrapani (Birmingham, UK), Dr. Kimberly Chapman (Children's National), Jill Franks (Propionic Acidemia Foundation), Dr. Andrea Gropman (Children's National), Dr. Eddie Island (Transplant Program, Georgetown University), Dr. Erin MacLeod (Children's National), Dr. Dietrich Matern (Mayo Clinics), Dr. Loren Pena (University of Illinois, Chicago), Brittany Smith (Propionic Acidemia Foundation), Kathy Stagni (Organic Acidemia Association), Dr. Marshall Summar (Children's National), Dr. Reid Sutton (Baylor Medical College), Keiko Ueda (Boston, Tufts University), Dr. Tiina Urv (National Institute of Health), and Dr. Charles Venditti (National Institute of Health). Dr. Gregory Enns (Stanford University) attended by phone and Dr's Matthias Baumgartner (University Children's Hospital, Zurich Switzerland) and Makoto Yoshino (Kurume University, Japan) are participating in manuscript preparation.

The manuscripts resulting from this meeting's discussions will be submitted for publication in the Journal Molecular Genetics and Metabolism.

Dr. Chapman and the sponsors would like to thank everyone who participated for their hard work and time.

## SIMD ANNUAL MEETING

Propionic Acidemia Foundation and The National Urea Cycle Disorders Foundation sponsored and exhibited at the 2011 Society for Inherited Metabolic Disorders (SIMD) 34th Annual Meeting in Pacific Grove, California. In addition to sharing information on propionic acidemia, we were able to network with doctors, researchers, dietitians, genetic counselors and members of industry. The meeting consisted of speakers and breaks to look at posters and exhibits. There were a number of posters this year relating to PA including case studies, gene therapy research, dietary management and complications.

PAF was represented by: (at right, l to r) Sharron Ames, Brittany Smith and Jill Chertow Franks



## PAF AWARDS STUDY EXTENSION

In October 2010 Propionic Acidemia Foundation awarded Dr. Jan Kraus of University of Colorado at Denver and Health Sciences an extension grant of \$28,000 for his study titled: "Genotype-phenotype correlations in propionic acidemia"

The project deals with the determination of the mutations or inherited changes in the DNA of propionic acidemia patients whose DNA samples have been submitted to the Corriell Institute. Knowing the mutations will lead to better understanding of the disease and lead to improved treatment for the affected patients. We will also determine which of the two mutations in each patient came from which parent.

***We want to hear from you! Have a PA story to tell, event to promote or news?  
Fall Newsletter submissions due by August 1!***

## CARBAGLU STUDY ADDS LOCATION

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

We intend to add Case Western Reserve University (CWRU), Cleveland, Ohio to the already existing sites at Children's National Medical Center, Washington D.C. and Children's Hospital of Philadelphia for our Carbaglu study. CWRU has received approval and they are ready to recruit for the study. Drs. Douglas Kerr and Laura Konczal will be the investigators at that site.

The recruitment poster for the CNMC in Washington, D.C. study is featured (right) on this page. Posters for the CWRU study will be forthcoming.

For more information about this study please contact PAF at 1-877-720-2192 or [paf@pafoundation.com](mailto:paf@pafoundation.com).



### NEW DRUG STUDY FOR PATIENTS WITH HYPERAMMONEMIA

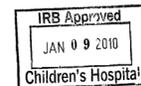
Researchers at Children's National Medical Center are looking for children and adults to participate in a NEW drug study for hyperammonemia, or elevated blood ammonia levels.

Eligible patients must be:

- Diagnosed with carbamyl phosphate synthetase I (CPSI) deficiency, N-acetylglutamate (NAGS) deficiency, Propionic Acidemia (PA), Methylmalonic Acidemia (MMA), or Ornithine Transcarbamylase deficiency (OTC)\*
- 1 to 70 years of age
- Willing to travel to Washington D.C. for a 4 day-long study and tests (travel and lodging paid by Children's National Medical Center)

If you, your child, or someone you know has any of the above diagnoses, you or they may be eligible for this study. Participation is voluntary and may help provide insight into an additional treatment option for children and adults with these conditions who suffer from hyperammonemia.

For more information, contact:  
Dr. Nicholas Ah Mew: [nahmew@cnmc.org](mailto:nahmew@cnmc.org)  
(202) 476-6177  
Children's Research Institute  
Children's National Medical Center  
Washington, DC 20010-2970



\*Patients with OTC deficiency must be female

## IN MEMORY OF LAUREN JUNE DELIMA



*Lauren June Delima*  
*June 14, 2001 - October 13, 2010*  
*Her life...celebrated!*

### Lauren's Purpose

Lauren was born with an inborn error of metabolism called Propionic Acidemia. Right from birth, she faced medical challenges, but Lauren was a true FIGHTER. She was fed a special low-protein formula via g-tube throughout the day and night. Lauren developed a heart condition called Dilated Cardiomyopathy before the age of 2. She was also diagnosed with Global Developmental Delay and Autism Spectrum Disorder at age 8. On May 21st, 2010 she was given the diagnosis of Congestive Heart Failure. The doctors placed her in palliative services and told us she did not have much time left. Due to her pre-existing medical issues, she was not eligible for heart transplant or devices. We were simply told to enjoy our last moments with her. We believe in GOD and His awesome powers. He gave Lauren strength for an additional 5 months to spend with her family and get acquainted with her newborn sister, Genelle. Although Lauren's medical status was grave, she did not show fear. Instead, she was determined not to al-

low her disease to control her, that to the last moments she was calm. We are in awe of our daughter's bravery! She was sentenced a life of hardship, but she went through it with total grace. She made an impact on everyone she came in contact with. Her purpose, in my unbiased opinion, is to show us all how to live life simply, yet beautifully. Lauren is an inspiration; a gift from GOD. "We love you deeply and your memories will continue to live in our hearts." Aubrey & Greg, proud parents.

<http://laurendelima.weebly.com>



## THE STORY OF ANDREW

Andrew was born on March 18, 1998. Everything went along quite well that day. Long labor and ultimately a C-Section brought a wonderful 8 lb. baby boy.

Andrew had been “sick” from birth. His doctor had no idea what was wrong. He exhibited many symptoms of a metabolic disease, but the medical community was just unable to “connect the dots” until the summer of 1998 when Dr. James Filiano from Dartmouth-Hitchcock in Lebanon, NH was able to diagnose Andrew with PA.

Initially, we had no idea what PA was and the horrible turn of events this dreaded disease would have on Andrew and our family.

Andrew went almost six months before diagnosis. Some were amazed he had gone so long without an “acute” incident of ketosis. However, the damage was still there. He had developed severe choric movements and was unable to move around like a six-month old baby. He had extreme low muscle tone and would projectile vomit on a daily basis. Looking back, these are all classic symptoms of PA.

Andrew started his “special PA diet” and it was around the same time he decided not to eat anymore, hence G-Tube surgery. For the most part, we learned to adapt to Andrew’s disease. Unfortunately, he was not able to walk independently and still uses a wheelchair or walker.

November 2009, we took Andrew to University Medical Center in Pittsburgh, PA for a Baclofen pump surgical install. This device was to help slow Andrew’s movements down to the point where he could possibly walk. Initially, the pump worked great. His movements were almost to a “normal” level. However, in late December 2008, Andrew developed acute spinal meningitis stemming from an infection of his Baclofen pump.

We rushed him to the hospital where they removed the pump and he was in the hospital for several weeks on antibiotic therapies. It was during this time Andrew developed severe cardiomyopathy. We feel that between the stress from the recent Baclofen pump surgery and subsequent infection, Andrew’s heart took “a beating”. It was also during this time we learned that children with PA are pre-disposed to heart issues.

His heart issues became worse and worse over time. So bad that we went to Children’s Hospital in Boston (one hour from where we live) to see if Andrew could get on the heart transplant list. Andrew was not found to be a good candidate for a heart transplant. The team felt that even if they performed a heart transplant, the “new” heart would run the risk of cardiomyopathy because he still has PA. With PA one is always more susceptible to heart issues.

It was at this point that Dr. Berry, Andrew’s metabolic doctor from Children’s Boston, proposed that Andrew get on the liver transplant list. Dr. Berry and his team stated that with a new liver and an active enzyme, Andrew’s health would greatly improve and there would be the chance other organs, including his heart, would “right itself”. Again, this was all speculation. There were no guarantees, but we had nothing to lose. We had to do something; otherwise, we knew Andrew’s chances of survival were slim.

In May 2009 at Children’s Hospital in Boston, Andrew had a liver transplant. During the first several weeks after surgery he experienced severe rejection, but with the exceptional medical care that was provided by Children’s Hospital staff, and lots of prayers, hope and faith, Andrew’s heart improved to the point he did not have cardiomyopathy... his heart was normal.

Since the transplant Andrew has been doing quite well. He no longer has any issues with PA. He is eating more and more by mouth and he seems to get stronger and stronger with time.

The movements are still there, but we are scheduled to see Dr. Duhaime at Mass General Hospital in Boston at the end of February in the hope that Andrew would benefit from a surgical procedure called Deep Brain Stimulation. We are hoping this procedure will stop the movements and Andrew might be able to walk independently.

For my husband, myself and our ten year old son, Jared, we want nothing else but to improve the quality of Andrew’s life. This has been quite a ride so far, we can’t wait to see what tomorrow will bring!!

Nicole J. Burnett

Mom to Andrew, age 12 and Jared, age 10



Andrew with the family dogs, Gretta & Boo

## CHARLOTTE'S STORY: FIRST BIRTHDAY!



On January 17, 2011 our darling daughter turned 1 year old. The past year has been the most exhausting, frightening, yet wonderful year of our lives. Charlotte has taught us so much and has brought so much joy to our family.

Charlotte is our first child. Like so many of the PA families, we noticed that something wasn't quite right with Charlotte just a few days after she was born. After our doctor expedited our newborn screening results and a subsequent confirmation of the PA diagnosis, our journey began. Over the past year, Charlotte has been hospitalized twice for metabolic acidosis.

The first hospitalization occurred a few days after her birth and the second hospitalization occurred when she was 9 months old. We have also made a few visits to the emergency room to flush elevated ketones from her system.

For the first 6 months of Charlotte's life, her amino acid levels and ammonia levels were stable and within the normal range. However, when Charlotte was about 7 months old, her ammonia levels became elevated and stayed elevated despite medical management. Of course, this just added to the stress and anxiety that my husband and I felt in trying to keep Charlotte healthy. Not only were we constantly concerned about acidosis and ketosis, but now we became more concerned with the effect that elevated ammonia levels may have on Charlotte's brain development.

In December 2010, two things happened that changed our lives. First, Charlotte started taking Carbaglu to decrease her ammonia levels. I was so thankful that my insurance agreed to cover this drug. In the few months that she has been taking Carbaglu, Charlotte talks and interacts with us so much more. As of today she says "mama", "dada", "dog" and "cat." But her favorite word is "cuckoo" (mimicking our cuckoo clock). It is the cutest thing I have ever seen.

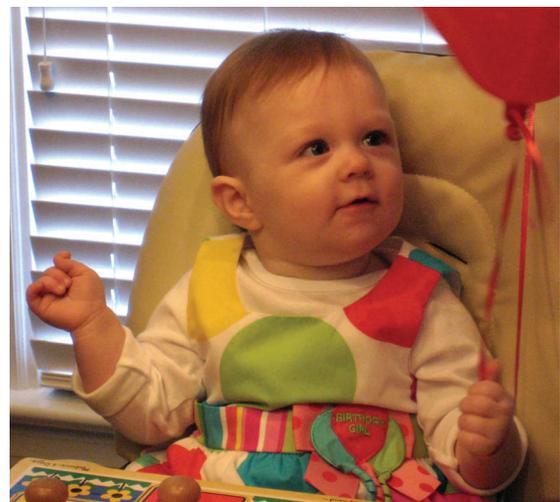
The second thing that changed our lives was our decision to get Charlotte a g-tube. The best advice I could give a family dealing with a new baby with a metabolic disorder is this: If you are going back and forth trying to decide whether or not to get your

child a g-tube...then it is time to get a g-tube. The mere fact that you are considering this type of surgery means that you have had trouble getting the required calories and fluid into your child. Trust your intuition - get the g-tube. After having the surgery, we have no problem making sure that Charlotte gets all of her nutrients and fluid. This surgery has increased our quality of life 10 fold. We are all able to get a full night's rest and we no longer have the added stress of constantly trying to get Charlotte to take her formula.

In preparation for Charlotte's first birthday, a day that we were not sure we would ever see, we wanted to do something special. In some ways her first birthday was a victory. As a family we made it through the toughest year we have ever had to go through. Having made it through the first year of this journey, we thought it was important to give back to all of the PA families. We downloaded donation cards from the PA Foundation website and sent them to our family and friends. We asked that in lieu of presents for Charlotte's first birthday, we would like for donations to be made to the PA Foundation. We plan to make this a birthday tradition. Not only will this increase awareness among our family and friends while raising money for a good cause, we hope it will also teach Charlotte how important it is to give back to a very important cause.

Growing up, I was taught that we have an obligation to give our time, talent and treasure to worthy causes. This maxim is now more important to me than ever before.

-Kathleen Cavanagh



*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](mailto: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

### EVENTS AND CAMPAIGNS

- Zumba for a Cure, Milford, CT- \$510 (top right)
- TGBK Shootout, Deerfield, IL- \$1500 (see cover)
- Adobe Cafe Bar & Grill, Philadelphia, PA- \$500
- Friends of Groucho's Sugar Bowl Party, Columbus, OH- \$1105
- Young Family Superbowl Party, King of Prussia, PA- \$1000
- Scrapbook for PAF, Columbus, OH- \$1140 (middle right)
- Thirty-One Charity Sale, Columbus, OH- \$200
- Tailgate Party & Corn Hole Tournament for PA, Gahanna, OH- \$16,000



### DONATIONS IN MEMORY- \$5000

- **Lauren Delima** (see pg. 3)
- **Bob Buck**- Gwen Mouat's granddad (bottom far right): *"Gwen's Granddad loved nothing more than spending time with his 3 grandchildren. He was always eager to help, and spent weeks sitting by Gwen's bedside & entertaining her during hospitalizations. He was very active in raising funds for PAF & supporting Gwen's family through all their trials. He was an amazing man. Not a day goes by that he isn't missed."*
- **Alice Dawe** - Chase Workman's grandma (bottom left): *"Chase's grandma loved him very much and Chase really enjoyed being with her. When Chase was born, Grandma Alice did everything she could to find out about PA and always wanted to help in any way. She would have been very honored that family and friends donated to this great cause in her memory."*



### DONATIONS IN HONOR OF

- Charlotte Cavanagh's 1st Birthday- \$275 (see pg. 5)



Join and follow  
Propionic Acidemia Foundation  
on Facebook

### SAVE THE DATE

- 7-30-11 Groucho's Poker Run, Columbus, OH
- 9-17-11 6th Annual Tailgate Party & Corn Hole Tournament for PAF, Gahanna, OH
- 10-16-11 Pounding the Pavement for PA:



Nationwide Better Health Columbus Half Marathon in Columbus, OH

Our Goal- Raise \$13,100!! \$1,000 for every mile of the race.

Join the PAF Pounding the Pavement for PA team or start your own!

Run in this race, a local race, around your block, or become a "virtual runner"

Contact Brittany Smith for more information at [paf@pafoundation.com](mailto:paf@pafoundation.com) or 877-720-2192

- **TBD- Mason's Salt River Lodge #180 Fishfry Fundraiser for PA, Mt. Washington, KY**

### Fundraise online by doing what you already do!

- ClubBing.com - play & donate tickets to PAF
- Search the internet through GoodSearch.com
- Shop through GoodBuy.com
- Sign up for eScrip at <http://escrip.com/>
- Sell items on Ebay using MissionFish. You pick what percentage goes to PAF.

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.

## PROPIONIC ACIDEMIA DVD PROJECT

### Propionic Acidemia project, a project reflecting the true meaning of giving back to the Propionic Acidemia Foundation



As child, I loved the story of "Stone Soup", which recounts how the power of persuasion is used to garner small contributions and this coming together and giving what you can improves the community soup for all in this tiny war torn village. In our story, the soup is a rich educational program for families affected by Propionic Acidemia and the villagers are wary Sponsors who see the

value of such an educational endeavor, but with a tough economy their funds are very scarce. So with a few dollars from Prevention Genetics and Vitaflo USA, we were able to film and edit a video of the first PA Education Day at Children's Memorial Hospital. The other "bits of carrots" as the story goes came from our others partners Hoffman Media and Dave Hildebrand Music, replication support from Datisis and artwork from Richard Koranda, of Two Advertising. Soon, our friends at Patient Power will be adding the program to our metabolic genetic disorders section of the patientpower.info.

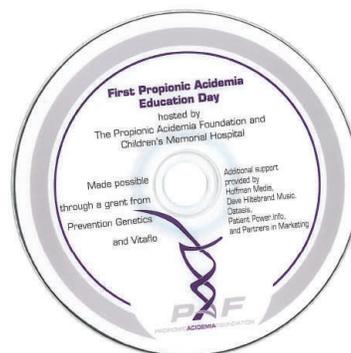
The PA patient education video presents top experts from Children's Memorial and UIC sharing the latest information on PA:  
 Chapter 1: "Introduction to Propionic Acidemia" and PA Families  
 Chapter 2: "Organic Acidemias" presented by Dr. Barbara K. Burton, Professor of Pediatrics at Northwestern University Medical School and clinical geneticist at Children's Memorial Hospital  
 Chapter 3: "The Genetics of Organic Acidemias" presented by Katherine H. Kim, MS, Genetic Counselor, Children's Memorial Hospital

Chapter 4: "Nutritional Aspects of Treatment" presented by Maryam Naziri, RD, LDN, Metabolic Dietitian, Children's Memorial Hospital

Chapter 5: "The Propionic Acidemia Questionnaire:"Survey of health status presented by Dr. Loren Pena, M.D., PhD, University of Illinois at Chicago

This patient program will be available on DVD as well as downloads from PA Foundation [www.pafoundation.com](http://www.pafoundation.com) and [www.patientpower.info/pafoundation](http://www.patientpower.info/pafoundation)

This educational program is good start but we need your support to continue our educational program in 2011 So just as *a bit of carrots* made the soup more delicious and rich; your donations will allow us to continue connecting you with top experts and sharing inspiring patient stories. If you are interested in sharing a DVD with someone you love or assisting us in our fund raising, donation to cover handling cost is \$5.00 for DVDs. There are a limited number of DVDs available.



Thanks again for your interest in Propionic Acidemia and our education resources. If you have ideas on fundraising of future topics please contact me.

Sharron Ames  
 Telephone 415.9393.2404  
[sharon@partnersinmarketing.info](mailto:sharon@partnersinmarketing.info)

### Help Us Find the Cure!

Name \_\_\_\_\_

Please send an acknowledgement to:

Address \_\_\_\_\_

Name \_\_\_\_\_

City, State, Zip \_\_\_\_\_

Address \_\_\_\_\_

Phone \_\_\_\_\_

City, State, Zip \_\_\_\_\_

E-mail \_\_\_\_\_

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

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PAF would like to thank  
Janice S. Boecker for her  
time and dedication.

We would like to acknowl-  
edge and thank each of you  
personally for making a  
difference for families affected  
by Propionic Acidemia.

Propionic Acidemia Founda-  
tion is run 100% by volun-  
teers and we couldn't do it  
without you.

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