

Propionic Acidemia Foundation

VOLUME 1, ISSUE 12

SPRING 2012

PAF Education Conference

Saturday, June 30th, 2012, 9am– 3pm

DDC Clinic-Center for Special Needs Children
14567 Madison Rd. (RT. 528)
Middlefield, OH 44062
RSVP to Jill at PAF by June 1st
Email: paf@pafoundation.com
Phone: 877-720-2192

Topics Include:
* Medications and Diet
* Clinical Trials
* Child Development for PA
* Cooking Demonstrations
* PAF Board Meeting
* Family Meeting Time



PAF Exhibits at SIMD Conference March 31 - April 3rd

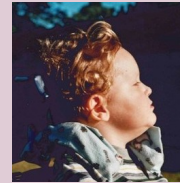
PAF exhibited and attended the 2012 annual meeting of the Society for Inherited Metabolic Disorders in Charlotte, North Carolina, from Saturday, March 31st to Tuesday, April 3rd, 2012. It was a great opportunity to listen to presentations, check out the poster session, and network with doctors, dietitians, researchers and vendors.

Congratulations!

Jen Mouat (mom to Gwen, 6 yrs, PA) was named the JPM Chase Central Ohio Volunteer of the Year! As a result of her recognition, JPM Chase awarded a \$500 grant to PAF in her honor. Thanks, Jen, for your commitment to volunteerism and service!



In Memory of Nicholas



July 24, 2001–
April 23, 2012
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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.

PAF AWARDS GRANTS

Marisa Cotrina, Ph.D. University of Rochester, Rochester, NY

"The impact of pa on brain astrocytes: an in vitro model to test mitochondrial therapy in PA" In January 2012, PAF awarded a \$27,000 grant.

Propionic Acidemia (PA) affects every cell in the body, but it has particular nasty effects in brain. Despite the profound damage that the disease causes to the nervous system, researchers have largely neglected to study how the brain manages the build-up of toxic compounds produced by PA patients.

Using genetically engineered mice to carry the same mutation that causes PA, our studies aim to decipher how brain cells react to the chronic effects of PA, particularly focusing on astrocytes. Astrocytes work as support cells for neurons; they eliminate metabolic waste and toxins (including ammonia) in brain and participate in the production of natural antioxidants. Astrocyte function is impaired in many neurodegenerative and neurological disorders, such as Alzheimer's disease, Parkinson's disease and Amyotrophic Lateral Sclerosis. These facts strongly argue for a possible role for astrocytes in the pathology of metabolic diseases, including PA.

In addition, as damage to mitochondria (the energy suppliers of the cells) seems to have a prominent role in a wide range of brain diseases, and propionic acidemia causes clear defects in mitochondrial function, we plan to test the efficacy of antioxidant reagents to improve survival and function of PA-affected astrocytes and, by extension, of brain function in PA patients.

Kimberly Chapman, M.D. Ph.D and Kristina Cusmano-Ozog, M.D., Children's National Medical Center, Washington, D.C.

"Is there energy deficiency in Propionic Acidemia". In January 2012, PAF awarded a \$15,500 grant.

Mitochondria are the powerhouses of our cells. Using a complex system, they act like little furnaces converting sugar, protein (amino acids) and fat into energy in the form of ATP. First, these molecules travel through the Krebs/tricarboxylic acid (TCA)/citric acid cycle generating precursors and energy holding molecules like NADH. These compounds are then fed into the oxidative phosphorylation pathway (electron transport chain) to generate ATP.

We think some of the complications seen in individuals with PA occur because they have difficulty making appropriate amounts of energy. This may happen for a variety of reasons. The block in propionyl CoA carboxylase (PCC, the enzyme that does not work in PA) may lead to the accumulation of toxins that don't allow other enzymes in the cell to work properly. Also, propionyl CoA is used to synthesize a portion of the body's succinyl CoA pool (an intermediate in the Krebs cycle). If PCC doesn't allow propionyl CoA to eventually be converted to succinyl CoA, then the Krebs cycle may not work as well as it should, decreasing the amount of ATP available for the cell to use. To determine if the Krebs cycle is dysfunctional in PA, we are using stable isotope labels to ascertain which components of the cycle accumulate in cell lines from individuals with PA as compared to cell lines from control individuals. This study (continued on pg. 7)

IN MEMORY OF NICHOLAS ALEXANDER PHILLIPS

July 24, 2001– April 23, 2012



Nick's buoyant attitude and gentle tenacity taught his family and friends how to love selflessly and carry on through hardships. His example will be a guide and comfort to those grieving his loss. He was a devoted aquarium enthusiast and played baseball with the Rivermont Dream League. He loved the River Park, his collection of movies and

books (especially Cars), horses, and his dog Buster. His parents are Kenneth and Sonia Phillips of Hixson.



PAF AWARDS GRANTS (CONTINUED)

Andrea Gropman, M.D., FAAP, FACMG, FAAN
Children's National Medical Center, Washington, D.C.

"Biomarkers for Neurological Injury in PA"

In January 2012, PAF awarded a \$27,000 grant.

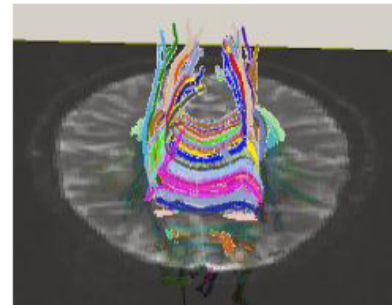


If you have Propionic acidemia and are between the ages of 7-60 years, you may be able to participate in an MRI study at Children's National Medical Center and Georgetown University.

Children's National and Georgetown are studying people with PA between the ages of 7 and 60 to investigate the differences between the brains of people with inborn errors of metabolism and healthy, typically developed people.

To be eligible to participate, you must:

- Be between the ages of 7-60 years old
- **Be medically able to undergo MRI safely**—no metal, such as braces, orthopedic devices, tattoos, or other items in the body
- Be able to follow instructions in English and have an MRI without need for medication for sedation (making one sleep)
- Be able to travel to Washington, D.C.
- **Be able to cooperate in the scanner without sedation for short periods of time**



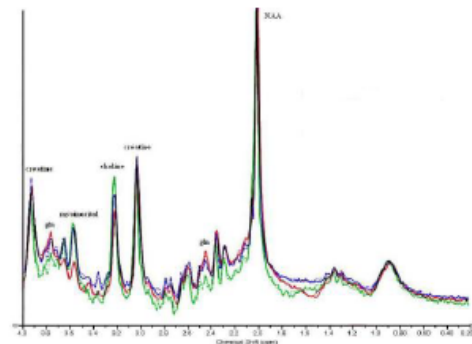
You are not eligible to participate if:

- You are pregnant or may become pregnant
- You have any metal in your body
- **Require sedation for MRI**
- You have a history of significant neuropsychiatric drug use

What does participation require?

- A series of imaging tests requiring about 3 hours, including an MRI (magnetic resonance imaging)

Compensation for time and travel provided.



Contact Information: To talk to a study coordinator and learn more about whether or not you are eligible to volunteer, please call the study team at (202) 476-3511.

Travel funding is available for participation.

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.

MAREN'S STORY

Hi, I am Honey Stecken. My husband is Scott. We have a 6 year old son, Lyric, and a new baby girl, Maren. Maren was born on February 13, 2012. She weighed 8 pounds and 6 1/2 ounces. Twelve days after having taken Maren home, on a Saturday, and while at our son's friend's birthday party, we received an urgent call from a doctor asking about Maren's wellbeing. We then learned she had an abnormal result on a newborn screening, elevated C3, which could indicate a metabolic disorder. A couple of days later, we received a call from a genetic counselor from Denver Children's Hospital who informed us that Maren was diagnosed with Propionic Acidemia. Maren shows no signs of her illness. She did not "crash" in the hospital and has not since. She is a month old now. Maren's little body was fed for almost 15 days a typical newborn diet of breast milk and formula. She had no abnormalities in her urine while having this. We feel so lucky she was able to hold her own. She now has a team of doctors and a special formula that includes carnitine, as well. Needless to say, as a family, we are devastated. We are slowly coming to grips with our new reality. She is a precious little girl and naturally we fear what's to come. We are still learning about PA. Looking at Maren one would never guess what's going on in her fragile little body. She appears to be just fine. As many other "seasoned" PA parents probably understand, we are in a pretty dark place having just been dealt this blow. We are scared, overwhelmed and shocked, slowly making our way towards acceptance and looking for the strength to persevere and smile while doing it. It feels like an impossibility right now. We are new to this community. We are willing to share our story and to hear from others.

-Honey Stecken



McKINLEY FISH FRY FUNDRAISER

The cities of Shepherdsville & Mt. Washington, KY really came together on Dec 4th raising money and awareness for the PA Foundation on Trent McKinley's behalf. The Salt River Masonic Lodge #1801849, with help from Mt. Washington Fire Dept Station 1, held a fish fry fundraising event that included music from the Branham family, karaoke, and a live auction. The event raised over \$6000! Trent's parents, Toni & Dave, were also interviewed live by a local radio channel educating the listeners about PA and inviting the public to the event. The local paper, *The Pioneer News*, also featured an in-depth story on spreading more awareness about

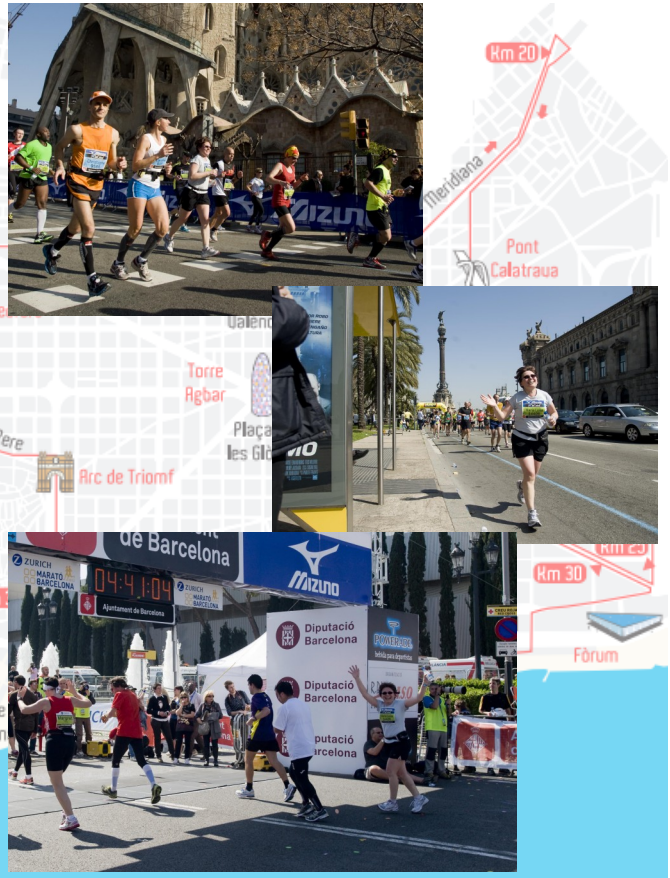
the disorder. It was a great week in Trenton's little town getting the word out on PA.



MARATHON BARCELONA FOR PAF

Dear All,
 We just got back from a wonderful trip to Barcelona. The race was amazing and it was truly special to run on the streets of my home town, with all my family cheering and my brother following me with his bike for most of the race. Thanks to the incredible generosity and support of friends and family, we raised \$11,095 for the Propionic

Acidemia Foundation, to support research on PA and, hopefully, find better treatments for all our children. For those who would like to know more about the race please I am including the stats of the race together with some pictures.
 Love, Marisa
 mom to Gabriel, 9.5 yrs, PA



Maria Luisa Cotrina
 These are your results:
 (total number of participants 19,400/13% women)

Real Time: 04:26:42
Official Time: 04:41:02
10 km time: 00:58:14
1/2 Marathon time: 02:03:45
30 km time: 03:00:07
Position: 13310
Category position: 588

GOLF TOURNAMENT FOR PAF

At 8:00 a.m. On August 31, 2011 the agents and staff of Re/Max Elite numbering 107 teed off at the Raven Crest Golf Course in Edmonton, Alberta, Canada, in support of one of our own. Peter Estephan, a well respected agent in our office has a daughter Alessia, inflicted with Propionic Acidemia.

The weather co-operated with a balmy 14 degrees Celsius. A fun time was had by all, and the day followed with a live auction. Many of the day's champions donated their winnings back to the office for donation to the Foundation. In all, \$6,336.84 was raised in honor of Alessia.



The Organizing Committee left to right:



Michael Pino, Kim LeClair, Peter Estephan, Paul Blais

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.

PAF FUNDRAISING SPOTLIGHT

EVENTS AND CAMPAIGNS

- 8/2011 **Re/Max Golf Outing**, Edmonton, Alberta, Canada, \$6336.84 (Canadian), (see pg. 5)
- 2/2011 **McKillop Golf Outing**, Binghamton, NY, \$1000
- 12/2011 **McKinley Fish Fry**, Mount Washington, KY, \$6000 (see pg. 4)
- 1/2012 **TGBK Shootout**, Deerfield, IL, \$2000 (below right and bottom right)
- 1/2012 **Young Family Superbowl Party**, King of Prussia, PA, \$1180
- 2/2012 **Scrapbook Crop**, Columbus, OH, \$1250 (bottom left and center)
- 4/2012 **Selvi Pragasam's Birthday Wish**, raised \$350! For my birthday on April 7th, I'm asking my friends and family for a special gift: help me raise \$250 for Propionic Acidemia Foundation! To donate online to Selvi's campaign and wish him a happy birthday: http://wishes.causes.com/wishes/457319?bws=creation_email_to_npo&m=313b961a

SAVE THE DATE

- 5/6/2012 **Cans for a Cure/Morgan Stanley Spaghetti Dinner & Silent Auction**, Binghamton, NY, Sandy McKillop (607-798-7924)
- 8/25/2012 **Groucho's Poker Run**, Columbus, OH, im2alesmom@yahoo.com
- 9/29/2012 **7th Annual Tailgate Party & Corn Hole Tournament**, Gahanna, OH, GwenForACure.com

Fundraise online by doing what you already do!

- Search the internet through GoodSearch.com: Goodsearch donations, \$1334
- Search or shop through iGive.com: iGive donations, \$3294
- 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



Join and follow
Propionic Acidemia
Foundation on Facebook

**We want to hear from you! Have a PA story to tell, event to promote or news?
Fall newsletter submissions due by July 31!**

FORMULA CAN UPCYCLE PROJECT

Michelle Ellis (Mom to Allison, age 6, PA) is an avid scrapbooker and crafter. All of those formula cans she was accumulating were bound to be converted into a crafty project! Best of all, it's an easy project that children and even non-crafty types can complete and enjoy!

Step 1: Cut scrapbook paper of choice to size for the can, leaving a half inch overlap to glue. Adhere to can with glue.

Step 2: Punch small holes in two opposite sides of the can at the top to make holes for the handle. Note: Michelle uses a heavy duty scrapbooking tool/hole punch for this.



Step 3: Thread a ribbon or pipe cleaner through the punched holes and attach by tying or twisting each end.

Step 4: Create a hang tag/mini card and tie onto the can at the point of the hole for the handle.

Step 5: Give to friends, family, co-workers, nurses, teachers or anyone else who might enjoy a unique and personal gift. It also makes a great "gift bag" for gifting small items, such as gift cards and socks. :-)



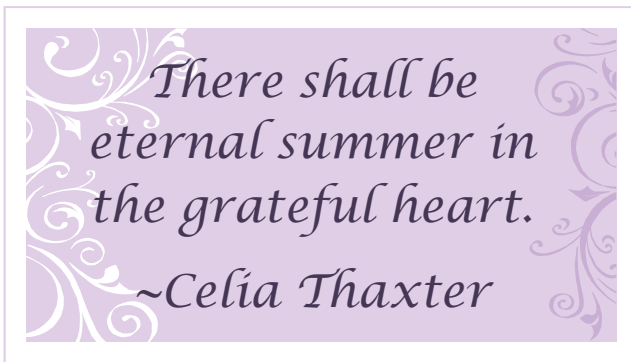
CHAPMAN STUDY UPDATE (CONT.)

(continued from page 2) will allow us to evaluate energy production in PA at the cellular level.

If energy is deficient on a cellular level, what does that mean for the person as a whole? A metabolic cart tool will allow us to measure resting metabolic rate or resting energy expenditure by indirect calorimetry. We are preparing a study to look at energy expenditure at rest when our patients with PA are healthy and when they are sick to see if this differs from other individuals who do not have PA.

We hope the information obtained from these studies will provide the foundation for more thoughtful treatment

approaches that will one day ameliorate the long term complications seen in individuals with PA.



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
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THANK YOU!
**Propionic Acidemia
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100% by volunteers
and we couldn't do
it without you.**

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