SEARCHING FOR A CURE HOPE FOR OUR CHILDREN

Propionic Acidemia Foundation

VOLUME 1, ISSUE 11 FALL 2011

Pounding the Pavement for PA Hits the Finish Line!

17 runners and walkers from Ohio, West Virginia and Illinois completed the Nationwide Columbus 1/2 Marathon on a beautiful October morning. We had a lofty goal of reaching $13.1K for this event and donations are at $14,006 and counting! We couldn't have done it without you! That amount includes $5481 raised at Maya's 5K in California (see pg. 6).

Join the PAF "Pounding the Pavement for PA" team or start your own! Run in any race, around your block, or become a "virtual runner". Contact PAF for more information at paf@pafoundation.com.

Families gathered at PA Family Day in Columbus, OH the day before the 1/2 Marathon.

In Memory of Aoun & Caitlin

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INSIDE

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

PAF Awards $32,000 Grant to Sarah Venezia and Jan Kraus at UCDHS.

Propionic acidemia (PA) is a disorder in which an enzyme known as propionyl CoA carboxylase (PCC) is mutated and does not perform its proper function. PA can be severe enough to cause death and mental retardation. Currently, there is no cure for PA and treatment is often severe. One possible treatment for PA is enzyme replacement therapy, where the mutated PCC enzyme would be replaced with a properly functioning one. This research proposal would like to look at a possible treatment for PA by this method. PCC is made up of two different subunits known as PCCα and PCCβ. People with PA generally have mutations in one or the other subunit, so we can deliver just the subunit that is mutated rather than the entire protein. Hurdles with enzyme replacement therapy include the delivery of the active enzyme into the patient cells as well as directing it to the correct location within the cell. In the case of PCC it needs to be delivered to the mitochondria. The mitochondria are separate membrane enclosed organelles within the cell that mainly supply the energy for cells. One promising way to deliver the PCC subunits across both the cell and mitochondrial embrane is the use of what is known as the TAT peptide. This peptide can cross cellular membranes and will also take along anything that is attached to it. Thus we propose to use the TAT peptide as a way to deliver the functioning PCC subunits to cells. In order to test this method as a possible treatment
for PA we will complete the following specific aims:

1. Attach the TAT peptide to each of the PCC subunits.
2. Test the PCCα or PCCβ TAT fusion proteins in patient cells to see if activity is restored.

PA Consensus Conference Update

Kimberly A. Chapman, M.D., Ph.D.
Children's National Medical Center

In January 2011, a group of clinician, metabolic dietitians, scientists, public policy specialists, and parental group representative including PAF's own Brittany Smith and Jill Franks met to discuss new recommendations for acute and chronic management of PA based on a summary of the natural history of PA and neurologic complications.

This has resulted in series of articles which will be published in Molecular Genetics and Metabolism in the near future and are available on line. The titles of the articles are as follows: "Acute management in propionic acidemia" (Chapman, et al.), "Chronic management and health supervision of individuals with propionic acidemia" (Sutton, et al.), "Natural history of propionic acidemia" (Pena, et al.), and "Neurologic considerations in propionic acidemia" (Schreiber, et al.). We are working with the publishers to be able to post them on this website. In the interim, ask your metabolic team to provide you a copy if you are interested.

NEWBORN SCREENING

Save Babies Through Screening Foundation Unveils New Website & Video

The new website: www.savebabies.org will serve as a comprehensive online resource center for families and health practitioners, and will host an interactive portal for new and expecting families to ask questions and view educational content about NBS, including SBTS's new video, "One Foot at a Time." Comprised of volunteers whose lives have been touched by NBS, Save Babies Through Screening Foundation is the only advocacy organization in the country dedicated to NBS. SBTS aims to educate parents, pediatric healthcare providers and policy makers about available comprehensive NBS, the importance of obtaining positive or other test results requiring follow-up actions within five days of birth and the importance of prompt confirmatory testing and treatment/management when needed. The Foundation's goal is to see that every baby born in the U.S. is screened successfully, effectively and comprehensively.

Genetic Alliance recently launched a Health Resources and Services Administration, HHS funded, newborn screening website, www.BabysFirstTest.org

The website is an objective resource for expecting and new parents to learn about newborn screening and is a place for families and health professionals to share their questions and experiences. Baby's First Test is a central site that brings together stakeholders and provides easy access to newborn screening resources from community groups, government agencies and public organizations. The site also features condition specific information, state information, family videos, and a Community Corner section, where visitors can learn about reliable sources of information pertaining to maternal and child health.

Preserving the Future of Newborn Screening (PFNS) *is a coalition of parents, health care and public health professionals, corporations and other individuals passionate about newborn screening. We hope to do our part to ensure better education about newborn screening, and about its benefits to society and the health of our children. We hope to educate the public about newborn screening and of improvements to public health through the use of blood spots left over from newborn screening. These blood spots are the "future" of newborn screening and must be preserved to save lives. PFNS operates as a project that is funded primarily through an educational consultancy with the National Newborn Screening and Genetics Resources Center (NNSGRC) in Austin, Texas.*

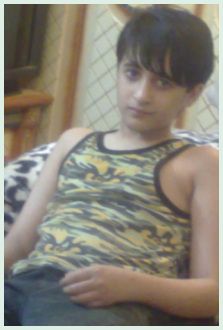
IN MEMORY OF CAITLIN & AOUN

Caitlin Waite
13 May 2005 - 12 April 2011

She was a very friendly little girl and touched so many peoples lives. We miss her so much. There was not one day the past 6 months that we have not thought about her.

Aoun
21 September 1995 - 19 March 2011

This is a short story of how we lost our son. He had cardio-respiratory arrest on 19th march 2011,which is a sudden attack. That night he had a minor fever, then he slept. He was alright at 7:30 am as his sister talked to him. We all were sleeping, then at 9:00 am we found him lying on the floor, we then brought him to hospital but he had died at home and we lost our precious diamond. We lost a very precious 15 and a half year old PA boy within half an hour.



PA NUTRITION GUIDELINES UPDATE

We wanted to provide you with a brief update on how far we have progressed with the development of nutritional guidelines for PA through our OA workgroup, a part of the Genetic Metabolic Dieticians International organization. We have analyzed and presented the results of our initial Delphi Survey where we asked a group of dietitians and doctors, with expertise in managing patients with inborn errors of metabolism, to complete a survey of approximately 90 questions on best practices for management of PA. We are currently working on setting up a meeting of a group of experts in a Nominal group meeting to discuss the results of the Delphi Survey to help determine our approach with developing treatment guidelines. Our workgroup has also been conducting an evidence-based analysis on all published literature in both the scientific, peer-reviewed journals, as well as non-peer-reviewed reports. We are extracting as much information from this literature as possible to help answer the 11 research questions that we developed for this condition. The answers to these questions will be used to develop the guidelines. Since the amount of evidence-based PA published literature is limited, the PA Delphi survey and Nominal Group meeting will help to supplement the information to be used for determining a consensus of 'best practices' from clinical experts in the field. We also plan to conduct a second Delphi survey to further clarify those areas where there is still some ambiguity on the best approaches for nutritional management. We are making steady progress and hope to have validated guidelines to share with clinicians by early next year. This has definitely taken a little longer than what we anticipated, but we are hoping that once our process is in place, we will be able to develop nutritional guidelines for other OA conditions more quickly. Our project is funded through a Human Research southeast regional collaborative grant.

Elaina Jurecki, MS, RD
GMDI and BioMarin

Keiko Ueda, MS, RD
Tufts Medical Center

ARCHIE'S STORY

Sat in a dentists waiting room opposite a young Mum and her son, when the dental nurse came in a called the Lady. As she stood up her son waved at me, his Mum looked at me and told me proudly "it's his latest thing" I immediately asked "How old is he?" She told me nine months.

I sat waiting for my turn and the only thought running through my head was, Archie is that age. Why isn't he waving?

This is what provoked us into seeking medical advice. We had appointment after appointment and were getting nowhere. We knew that things were not right. He was too good, never crying, not really paying attention, barely sitting let alone waving.

The only answer we got for a long time was "it's delay" this is an official medical term in the UK for "we haven't got a clue!"

After many months and continual pushing we finally got an answer…"Autism" or rather Autistic Spectrum Disorder. Again an official term that means "covering ours butts because we still haven't got a clue"

Finally after 18 months and many consultations and Archie developing a curious game of in and out where he would put items in something and then remove them. It didn't matter what he used from toys cars in a box to video tapes from the shelf. The game was the same. We went for a blood & urine test, the results from this takes a week. After a week we were called in again for the same thing. The following day I received a call from the Doctor telling me to bring Archie to the hospital immediately. Now worried sick and fearing God only knows we went. This is when we were told of the PA. At first I didn't believe there could be such a problem, after all protein is in just about everything surely?

The condition was explained along with the odds of having a child with this condition. A single diet sheet was provided and the powers that be went to great length to explain that the amount of protein he could have was worked out on a weight ratio. When I questioned this along with many other things it boiled down to "it was their best guess".

Not long after, Archie now being 30 months old, he had an eye test. It was found that he needed pretty strong glasses as well. When I asked if it was part of the condition the reply was no it's just more bad luck. To our amazement Archie never tried to remove them, therefore in my mind the only reason for this was for the first time in his short life he could actually see!

Now eight years on and too many scares and hospital visits to mention Archie is your average (albeit small) 10 year old who knows he is different. I know it gets him down on occasion and he knows he gets treated differently from other kids, not always for the better as it can lead to him being left out or ignored. He does his best and that's good enough for me.

Dave Koncher

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.*

JULIEN'S STORY

Our story with PA begins with a phone call on our son's third day of life. Up to that point things had been relatively smooth. It had been a year and a half since we began the process of adopting a child, and a little more than two months since we met with a birth mom who chose us as the adoptive family for her son. The joy we felt when we finally held that beautiful baby boy in our arms for the first time was indescribable. Things were like a whirlwind - emotions, social workers and family phone calls all passed in a blur. There were a few warning signs: he was hypothermic and gagged at almost every feed, but these things were explained as "normal" by the nurses and our minds were set at ease.

Early on the third day I received a phone call from our adoption case worker that overnight Julien had been taken to the NICU as a precautionary measure because the overnight nurse had seen some things that caused concern. Throughout the day things only got worse. With each new phone call we learned about a new piece of what was being done to care for our child and nothing sounded good. By the time we arrived at the hospital late that afternoon Julien had been in the NICU for more than 12 hours. He had been intubated because of respiratory distress and was hooked up to more medical equipment than seemed possible with a body as small as his. I'll never forget the NICU doc saying to us as we met late that night that they suspected a metabolic deficiency, and that depending on which one it was he would not be able survive.

Julien was transferred to Children's Hospital that night where the genetics team handling his blood work was stationed. Over the next ten days we alternated with the birth mother to make sure that someone was there for J almost all the time as we waited for a diagnosis and a plan to care for him. Propionic Acidemia was the second diagnosis we were given. I remember being grateful for this because PA was much easier to pronounce than what they suspected initially. Long days in the NICU and long nights at home wishing we were with him in the hospital were spent reading website after website as well as hospital printouts to learn everything we could about PA and its effects on the children who are born with this condition. Knowledge felt empowering and overwhelming at the same time.

Julien was released from the hospital 10 days after his birth. We were once again filled with joy to be finally coming home - all 3 of us - and anxious about trying to manage him while feeling alone. We were first time parents, and now none of the stuff we had read about what to expect applied. We followed the plan given to us by our doctors, worked to make sure that the schedule of feeds provided by our nutritionists was implemented and prepared for the inevitable fatigue of the months in front of us.

Julien managed the first year fairly well. He was a great eater at first and his care was relatively easy. Other than having to follow the carefully regimented schedule (even overnight) we charted everything that went in and everything that came out. We tracked meds, supplements, formula and attitude. We met with our genetics team each month, giving careful updates and watching for any warning signs. We met with a pediatric cardiologist and established baselines for J so that we would have a standard by which to check him should anything come up in the future. We did have a few hospital stays as he battled colds and other illnesses that brought on metabolic crises. Twice he came home with an NG tube so that we could continue to manage his feeds and meds until his appetite returned.

Over time his appetite diminished to the point that we proceeded with a G tube placement at 11 months. We also participated in a research study to test Carbaglu and whether or not it would help keep J's ammonias down to a level that was closer to normal. Some of our conversations with doctors have been quite unnerving - like when we talk about the possibility of a liver transplant. Some of them are very encouraging as we track his development and learn more about how his body is able to handle the effects of PA.

For now Julien is a relatively healthy, very happy 15 month old little boy. He receives Early Intervention services (OT, PT, and DT) and he is testing about 3 months delayed. He has in incredibly goofy sense of humor, loves to cruise around the house, explore whatever sits below a three foot height and babbles constantly. He's never met a person he didn't like and is a constant source of joy. We are so proud of him - not just as our son, but also because of the incredible battle he has waged since those first couple of days. When we picked Julien's name we did so because of the meaning behind it. When you put J's first and middle names together you get the meaning of "young warrior." We chose this for him because as a transracially adopted child we knew he would face some tough circumstances. We had no idea the physical battle he was going to face, but he has proven himself worthy of his name time and time again.

We cannot fully express the joy we feel to have Julien as our son. The privilege of adoption is an incredible one and no medical diagnosis can change that. We believe that God is working his plan for our lives through all of this, and even when it is not exactly the way we would choose if we could, we are grateful that he brought Julien to us and we're excited about the life ahead of us.

PAF FUNDRAISING SPOTLIGHT

EVENTS AND CAMPAIGNS

- 3/2011 **Luck of the Irish Challenge**, One National Foundation, Michigan, $2700, Marody Family
- 3/2011 **McKinley Basketball Fundraiser**, Shepherdsville, Kentucky, $450
- 4/7/11 **Selvi Pragasam's Birthday Wish on Facebook Causes:** Got to celebrate my birthday with a heart filled with love and support from friends who immediately heeded to my Birthday wish on Facebook and supported me in raising $230 for PAF. God bless all those who donated and hope there will be a cure soon for the sake of all our children.
- 4/30/11 **3rd Neighborhood Beautification Day**, New Albany, OH, $2464. Energetic volunteers painted or replaced mailboxes in the neighborhood in exchange for donations to PAF.
- 6/2011 **Cans for a Cure**, Johnson City, NY, The Community Based Education Can Collection Team gathered cans and bottles throughout the school year and raised $335.00 for PAF in honor of Connor McKillop. The project allowed students to practice sorting, counting and money skills as they prepared the recyclables for redemption.
- 8/13/11 **Groucho's Ride for PA**, Columbus, OH, $3800
- 8/14/11 **Maya's 5K**, Vasona Park, CA, $5481– as a part of Pounding the Pavement for PA (right)
- 9/17/11 **6th Annual Tailgate Party & Corn Hole Tournament**, Gahanna, OH, $11,471 and counting!
- 10/13/11 **Lauren's anniversary fundraiser,** Vancouver, BC. (below) We raised $2180 from selling plants propagated in her memorial garden. Our family/friends provided the food, the weather cooperated and attendance was amazing! We are so blessed and thankful.
- 10/16/11 **Columbus Half Marathon**, Columbus, OH, $8525 and counting! (see cover)

Fundraise online by doing what you already do!

- Search the internet through GoodSearch.com: Goodsearch donations, $1258
- Facebook Causes: $655
- Search or shop through iGive.com: iGive donations, $3098
- 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? Spring newsletter submissions due by March 30!

PAF FY 2010-2011 ANNUAL REPORT

FINANCIAL REPORT
Revenue:
Contributions: $80,236
Interest Income: $496
In Kind Donations: $1,888
Total Revenue: $82,620

Expenses:
Research Grants/Exp.Grants: $28,000
Programs & Outreach Total: $5,417
Printing (in-kind): $1,888
Fundraising Total: $751
Operational Expenses: $1,486
Total Expenses: $37,542

Cash Assets 8/1/2010: $81,648
Cash Assets 7/31/2011: $126,726

BOARD OF DIRECTORS/OFFICERS
Jill Chertow Franks, President
Brittany Smith, Secretary/Treasurer
Ann Marie Young
Michael Rosenson

Board disclosure: Jill Chertow Franks is the sister-in-law of Michael Rosenson.
Donations made by board members totaled $929.

PROGRAM ACCOMPLISHMENTS
Research:
Participated in the first PA Consensus Treatment Conference, Washington, D.C.
Grant Disbursement:
$28,000 - Jan P. Kraus, Ph.D., CDHSC Denver, CO: "Genotype –phenotype correlations in PA"

Outreach:
- Published and distributed fall and spring newsletters to affected families, clinicians, and donors
- Exhibited and attended Society of Inherited Metabolic Disorders Annual Meeting
- Held PAF Education Conference in conjunction with Children's Memorial Hospital, Chicago, IL
  -Video of the conference is at: www.pafoundation.com
  -Presented at Homocystinuria Conference, CDHSC, Denver, CO

We would like to acknowledge and thank each of you personally for making a difference for families affected by propionic acidemia.

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.

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