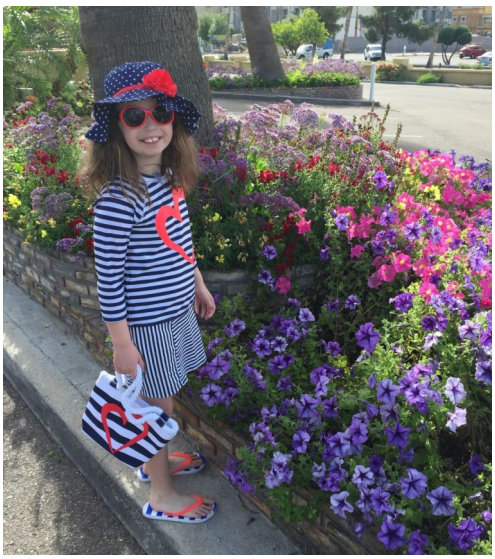
SEARCHING FOR A CURE HOPE FOR OUR CHILDREN

Propionic Acidemia Foundation

VOLUME 1, ISSUE 18 SPRING 2015

Family Story Issue

GWEN'S STORY



My beautiful girl just turned 9 years old this year and it seems nothing short of a miracle. At 2 days of age, Gwen became catastrophically ill, her body temperature dropped below 90 degrees, ammonia level exceeded 1,500 umol/L and she stopped breathing. She was placed on a ventilator and received peritoneal dialysis for a couple of days until she came out of her coma and was breathing on her own. On her 3rd day she was diagnosed Propionic Acidemia and her future was very uncertain. During Gwen's first 3 years of life she spent as much time in the hospital as she did at home.

Although she's been admitted more than 50 times, she's undoubtedly one of the happiest people on Earth. At age 1 she stopped eating by mouth, and since then she's been fed 100% by a feeding tube because she refuses to eat anything. For many years she wore a backpack to carry her feeding pump, but she is now able to tolerate her formula through small bolus feedings and has a nurse who cares for her during the day.

Gwen knows she's very cute and she plays that to her advantage. What she does not yet know is that she's very brave, has an endless capacity to forgive, an amazing will to live, and a beautiful spirit from God that has touched the lives of hundreds. She talks non-stop, sings the entire time we're in the car, jumps off of anything she can climb on, loves to dance, play with her American Girl dolls and spend time with her brother and friends. She's in second grade and receives special education services for PT, OT, math and reading. She's also in Brownies and on the Special Olympics swim team! She is a miracle, a daily blessing, and a ray of sunshine in any room. I am grateful for every day I have with her and so proud to be her mom. Jen Mouat jenmouat@gmail.com

PA International Patient Registry

Help move research forward for propionic acidemia. Participate in the Propionic Acidemia International Patient Registry.

As of April 1st, there are 38 participants. For more information on joining the registry or to update your information, go to www.paregistry.com

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 CONTINUATION GRANT TO LOREN PENA, MD. PH.D, DUKE UNIVERSITY

Update on "Laboratory parameters reflective of metabolic control in individuals with propionic acidemia" at Duke University

Understanding how the results of laboratory tests relate to a person's current health, treatment options, and future health risks can be invaluable. However, this is an area on which little information for people with propionic acidemia (PA) is available. To address this question, we are measuring and comparing levels of plasma and urine metabolites in people with PA when they are well and during illness. By doing so, we hope to identify laboratory tests that can help healthcare providers decide on the best available treatments, and identify patients most at risk for developing health issues such as pancreatitis.

Since the research study began in April 2013, we have received samples from 11 participants. Participants provide urine and blood samples for the research study during regular visits to their metabolic specialist and if they are hospitalized while ill. We are also including information from samples previously processed at Duke, and reviewing medical records and laboratory test results from the participant's treating physician.

We have already seen some promising results that warrant further investigation. This includes:

Differences between the values of specific amino acids found by comparison of amino acid levels in approximately 110 samples from well individuals with those in 20 samples obtained during illness. We will continue to focus on these amino acids during analysis of future samples.

Results from our analysis of urine organic acids suggest dysfunction of the tricarboxylic acid (Krebs) cycle, a series of biochemical reactions that produce ATP, the energy currency of the cell. These results confirm previous findings. Continued investigation may help to determine whether treatment with metabolites in the TCA cycle *could* be helpful.

As part of the study, fatty acids (components of fat molecules) were measured in blood samples in a small number of participants. Our exploratory data warrants further investigation of odd chain fatty acids as long-term markers of metabolic control.

We still have a wealth of data to analyze and will continue to enroll new participants and collect samples now that the study has entered its third year. We greatly appreciate the support of this study and would like to thank all of the families who have contacted us. For questions about the study, please contact the study coordinator, Jennifer Goldstein, at phone number (919) 684-0626 or email jennifer.goldstein@duke.edu

PAF AWARDS A NEW GRANT TO MICHAEL A. BARRY, PH.D, MAYO CLINIC COLLEGE OF MEDICINE FOR "NEUROLOGIC PHENOTYPES AND THERAPY IN PROPIONIC ACIDEMIA MICE"

You can read more about Dr. Barry's research in our Fall 2014 newsletter or see the following papers published last year:

Long-term Sex-Biased Correction of Circulating Propionic Acidemia Disease Markers by Adeno-Associated Virus Vectors.
Guenzel A, Collard R, Kraus JP, Matern D, **Barry MA**.
Hum Gene Ther. 2015 Feb 5. [Epub ahead of print]
PMID: 25654275 [PubMed - as supplied by publisher]

Effects of adeno-associated virus serotype and tissue-specific expression on circulating biomarkers of propionic acidemia.
Guenzel AJ, Hillestad ML, Matern D, **Barry MA**.
Hum Gene Ther. 2014 Sep;25(9):837-43. doi: 10.1089/hum.2014.012. Epub 2014 Aug 21.
PMID: 25046265 [PubMed - in process]

Organ or Tissue Donation– Biorepositories

Organ or tissue donation is a priceless gift that facilitates research into the disease process and promising new treatments for any disease. Although we may frequently think of organ donation as a life-saving procedure for the recipient, donation is also a valuable resource for medical research. In rare disorders, donation of organs or tissue provides an extraordinary opportunity for microscopic, molecular, and biochemical examination to gain insight into the how and the why of clinical symptoms observed during an individual's life. In addition, donated organs and tissues can be used to investigate promising new treatments for the disorder. As an example, pre-clinical studies of safety and toxicity of a candidate treatment are frequently undertaken in model organisms. Tissue derived from humans affected with the disease in question can become an additional resource for pre-clinical studies.

Donated organs and tissues are stored at a designated location, known as a biorepository or a biobank. The PAF's initiative of storage of tissue through the Coriell Institute for Medical Research is one example of a biorepository. Biorepositories may be part of a research protocol, and inclusion of the organ or tissue requires informed consent from the individual or his/her guardian. Donation into a biorepository therefore requires prior planning and consideration. The sample may become available upon the individual's death, or on receipt of an organ transplant. Samples receive an individual identifier, and basic information about the individual is included, along with clinical information such as the specific disorder and medical history. Samples are available to investigators, and there is usually a specified mechanism for evaluating a request for tissue.

Families may seek the opportunity of a positive outlet to the grief associated with a loved one's passing through organ donation. Your medical provider can be a valuable resource for questions, concerns, and potential opportunities. Additionally, research studies may be listed at http://www.clinicaltrials.gov.

Please contact me at loren.pena@duke.edu if you have any questions.

Loren Pena, MD, PhD, Duke University

Evan's Story

Evan is now 8 years old. He had a partial liver transplant in March 2012. Whilst the 1st years was very tough, unfortunately he was diagnosed 6 months after transplant with Lymphproliferative Disease. A type of Lymphoma brought on by the immunosuppressants needed to stop his body from rejecting his new liver. He needed a few months of chemotherapy but thankfully got the all clear from this a few months later. Since this however, Evan has come along so so much.

The transplant wasn't a 'cure' but is a huge help in managing Evan's condition and giving the best chance at living a normal life. He can tolerate a lot more protein which in turn helps his development and generally makes him look healthier. He is in his 2nd year in the local special needs school and he absolutely loves it. He runs out to the bus in the morning and always has a smile on his face wen he gets off the bus when he gets home! He loves the attention and being with others his own age. His speech and communication is coming along although we still cant converse with him and his eating is steadily improving. Although foods still have to be pureed, he is open to trying new things and experimenting and playing with food whereas before he showed absolutely no interest in food and mealtimes.

He is still tube fed most of the time via a mic-key button in his stomach. His feed consists of Energivits, Nutrini and MMA/PA Gel and all medications we give to him via this tube. So even though he still picks up every bug and infection going and does have long bouts out of school and the odd admission to our local hospital the future is looking bright for Evan.

There is no doubt life is tough living with PA, we do our best by our children. Not everyone agrees with our decision to go for a Liver Transplant but even though the 1st year was tough we don't regret our decision. Our thinking was that if something happened during or after the transplant at least we are doing everything possible and in our power to give our child the best chance possible to have a normal life. Life with PA is so unpredictable.

Sarah (Evans mam)

ALLISON'S STORY

Allison Ellis, Propionic Acidemia, age 9

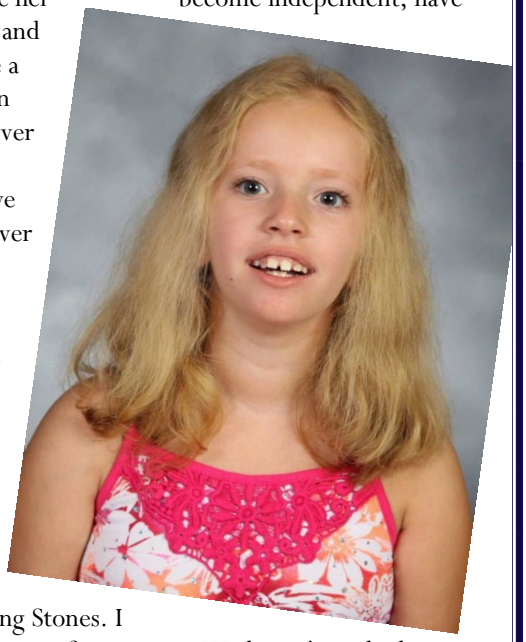
I can't remember the last time I wrote an article or update about my amazing girl…I also can't remember the last time she was in the hospital before now. It is Friday, March 20th, and she was admitted the evening of the 17th. Thankfully, hospital visits are less frequent the the first few years of her life. We are here now because she has a stomach bug like the rest of the family, but couldn't get better on her own like we did. So…IV fluids (D10), Carbaglu, and "sick day" formula…

Potty training has been one of our biggest challenges. We have tried lots of things including the potty watch, and just not using pull-ups, but underwear instead. Very messy! The combination of her diet, delays, and ulcerative colitis make it necessary for us to use chucks on her bed every night as well. I just paused and wandered off thinking about whether or not she gets teased or made fun of at school. I am back now because I don't want to think about that. I know I wasn't always a nice kid, especially when I thought someone wasn't "normal".

Nationwide Children's Hospital is an incredible place, with an amazing staff. Before I go on about Allison, I would like to thank some of the most special people in her life. "Daddy" Dave, who does EVERYTHING! Grandma Char, who cares for her and/or her twin brother Austin for countless hours whenever needed. Yvette Williams, her home nurse for over half her life, and full-time nurse at Nationwide Children's Hospital for 20 years this August! Dr. Bartholomew, Jimia Hoy, and everyone in the Genetics department, and all of the wonderful nurses on the 11th floor (many who have cared for Allison since she was a week old!) We love you all and couldn't do this without you!

Allison is currently in 3rd grade and is in a special needs class (another special person…Kelly Duell, intervention specialist who has taught her SO much, every year since kindergarten!) Despite her developmental delays, she is reading and writing many words, doing math on a calculator, and expressing herself more than ever! She wears DAFOs, receives physical, occupational and speech therapy weekly. She loves music (Frozen soundtrack, Laurie Berkner, and nursery rhymes, mostly), dancing, playing hide and seek, arts and crafts, swimming, and playing with her friend Gwen. There isn't much she doesn't like, all I can think of is storms, dogs (but she is getting better), and she isn't very interested in food. Primarily G-tube fed her special formula (which consists of Propimex 2, whole milk, MCT oil, water, and levocarnitine) she is only allowed 3-4 grams of protein a day by mouth. She will snack on her favorite…"white cookies" (mini vanilla Oreos introduced by Grandma Char), and occasionally have a little applesauce, juice, or taste of something (but when asked usually replies "no, I don't like that anymore").

Our goal for Allison is to be as "normal" as possible. We would love to see her become independent, have a job she enjoys, and maybe even have a family of her own someday. Whatever she wants is our wish, and we have to help her discover what that is.



Michelle Ellis
Columbus, Ohio

P.S. (Just for fun)…I used to joke that Allison's "Make-A-Wish" was to meet Mick Jagger and the Rolling Stones. I even got her to say it a few times :) We haven't applied for anything like this for her, because we really aren't sure what she would pick. I just asked asked her what she most wanted, what she wished for…she answered "I wish for Allison". I just love her! I wish for a cure for Propionic Acidemia and will continue to do my part fundraising and supporting the wonderful PA Foundation.

I wish to meet Mick Jagger, so if anyone reading this has any connections… email im2alesmom@yahoo.com or find me on Facebook as Michelle Mickee Orcutt Ellis. LOL

NALANI'S STORY

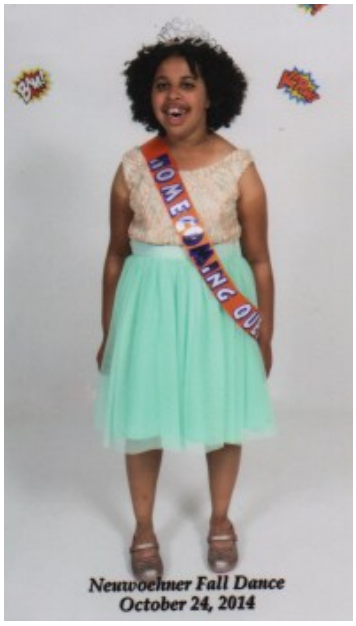
My daughter Nalani is in High School!! Her middle school educational experience was the toughest we have had thus far. The IEP's were giving me panic attacks. Nalani was getting more and more withdrawn and unhappy. I had to do a lot of research and soul searching to decide what to about high school. We live in a really good school district and I am aware of the importance of inclusion but, after visiting one of the special school districts self-contained schools I decided we would give it a try. Nalani has not attended one of the special school district schools since preschool age 3-5.

From the start of school she seemed a lot happier. There were some bumps in the beginning but, nothing major. The school is awesome, she is able to do things that she would not have been able to do in the district high school, but is still allowed to participate in "Sparkle" a district inclusive cheerleading program after school at the public high school.

Her class consists of a total of 6 students, 2 aides and 1 teacher. The students are put in classes according to their abilities. Her new school has lots of family activities and so far 2 school dances one of which they crowned her and a few of the other new girls homecoming queen. They offer a "bootcamp" activity where the students at all levels work together to perform tasks, she loves this. Her teacher writes me notes everyday about what she does at school and sends pictures of special activities. I send her pictures of activities she has done over the weekend and they discuss them at school on Monday. She gets books and educational material to help at home. They check on her when she is sick, which may not seem like a big deal but, it really means a lot to me.

She has been there a little over 6 and a half months and just had her first IEP. They changed a majority of the things I had been asking to be changed at the middle school in her IEP before I even had to ask. The speech therapist is so amazing that after 10 years we have stopped paying out of pocket for a private speech therapist. During the IEP they brought her in and had a PowerPoint presentation, including pictures of some activities she had done. The staff went around the room and everyone said something positive about Nalani. She really loved this, and was so incredibly proud it brought tears to my eyes. For the first time in years I enjoyed her IEP and did not feel like I was going into battle, I felt more like it was a group of people that cared about Nalani working together on a plan for her.

I always hear about advocates of inclusion and being in the least restrictive environment. Someone said to me when I made the decision to pull her out of public school where she was included in general education classrooms, that she needs to know what it is like in "real life situations". I still worry about this but, when I look at the new confidence that Nalani has I believe I have made the right choice for her. I know that each child and family has a unique set of educational needs, I just wanted to share my experience with a self-contained school, and share a photo of my homecoming queen!



Neuwoehner Fall Dance
October 24, 2014

Angela Waits
Mother of
Nalani age 15 PA
Xavier age 18 months

NEW FORMULAS FROM NUTRICIA!

At Nutricia, we are committed to providing advanced medical nutrition to ALL patients with inborn errors of metabolism, including those with Propionic Acidemia. We are proud to announce the upcoming launch of our newest Methylmalonic Acidemia & Propionic Acidemia formulas for infants to juniors and adults! Introducing the **new** *MMA/PA Anamix® Early Years* for infants up to 12 months of age, **and** *MMA/PA Anamix® Next* for ages 1 year and older.

Following are a few key features of the new MMA/PA Anamix formulas:Contains prebiotic fiber, to help support the digestive system:

- Contains DHA & ARA; DHA is important for brain and eye development in infants and children up to two years of age
- Contains Calcium and Vitamin D to meet current guidelines and to help build and maintain strong bones and teeth.

The MMA/PA Anamix Early Years will be available in June. Talk to your dietitian for more information or contact nutritionservices@nutricia.com or call us (800) 365-7354 Monday – Friday 8:30am – 5:00pm

PAF EVENT & FUNDRAISING SPOTLIGHT

UPCOMING EVENTS AND CAMPAIGNS

- **CH Tourney, Columbus, OH– October 2015**

PAST EVENTS AND CAMPAIGNS

- *Summer 2014*- **Amanda Matz Multi-family Garage Sale,** Washington, $3,000
- *January 2015*- **Branch Family Superbowl Fundraiser,** Kokomo, Indiana, $500
- *March 2015*- **Hoops for a PA Cure - McKinley March Madness Brackets,** Kentucky

ONGOING CAMPAIGNS

- **Igive**- $4,129.42 Total (Searching: $27.92 Shopping: $3,896.50 Bonuses: $205.00 Number of supporters: 112 Please share information with family and friends about Igive!!!
- **Annual Appeal**– Goal $15,000!
- **Goodsearch**- $1409.11 (including Goodsearch, Goodshop, Gooddining, Goodgames, Goodsurveys, Goodoffers, Goodtvads and Goodswipe) Number of supporters: 342
- **AmazonSmile -** $60.45

DEDICATED GIFTS FROM INDIVIDUALS

Among the many contributions from individuals received, the following is a list of some that were dedicated to those who have inspired the giver.

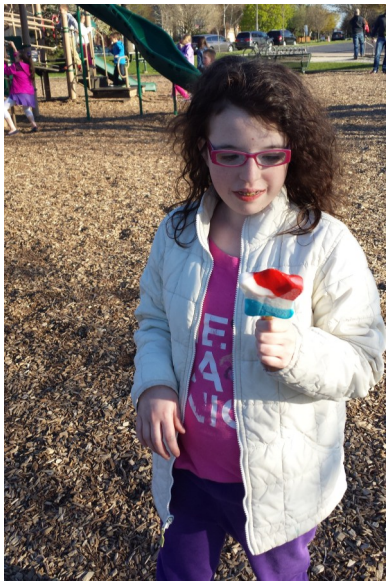
- **Gifts in honor of:** Kristin Boecker, Jordan Franks, Varun and Maya Gattupalli, Dylan Jaehnke, Nalani Johnson, The Lenerts, Edgar Jr. and Dania Martinez, Zachary Matz, Maren Stecken, Benjamin Sweetman, Brett Young

- **Gifts in memory of:** Alice Dawe, John Dawe, Sharon Esses, Vincent Philip Franze, John R. Hatcher, Timothy Huff

Thank you for all donations and the kind notes we receive throughout the year. Your support overwhelms us and continues to be a source of inspiration. PAF couldn't do what we do without your incredible support.

TALLI'S STORY

Talli is seemingly always on the go, when she isn't she wants to know what the plans are for after school and the weekend. She loves to go to school and hates missing class if she is sick or has an appointment. She can't stop talking about an upcoming trip to see her grandparents in Oregon and a sleep-away camp with the teenage girls from our Church over the summer. She loves technology, music, and live performances. We have had to put on parental controls on her tablet just like her typical peers have. She got braces on just after her birthday in the fall.



She is in the 7th grade at our local Junior High School. She is in an Instructional class for her core classes and is mainstreamed for PE, science, choir, and a period that rotates between art, technology/engineering, sewing, and cooking. This year she was on the school's 7th grade cheerleading squad and she just excelled! She attended practice every day and then came home and practiced some more. She is very excited for ext season.

Over time, new health challenges have come up and she now has some complicated treatments. She was fortunately newborn screened; we found out a week after she was born. She was already in crisis at the time and took several weeks to stabilize her in the NICU.
(continued page 7)

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.*

TALLI (CONT. FROM PG. 6)

The biggest challenge at that time in keeping her stable was severe food allergies and reflux. She vomited every feeding! Thankfully, she has outgrown some of her food allergies, however, tree nuts and latex can afect her by just being around them.

She had frequent infections and was hospitalized every couple weeks when she was an infant and toddler. After starting IV immunoglobulins (IVIG), she had a significant reduction in the number of illnesses and hospitalizations. She has had sepsis several times, one of those times infecting her hip which required surgery. She has a significant Long QTc wave and had an internal defibrillator placed when she was in the 2nd grade. She also has hypothyroidism and was recently diagnosed with growth hormone deficiency, so takes daily growth hormone injections.

After many years of going to the hospital for IVIG infusions, she will be starting weekly subcutaneous at home. This will keep her better protected from infections, but also free up her (and our) schedule and cause fewer side effects. She had received IVIG at our local hospital which has a Pediatric Critical Care Unit because of poor tolerance to it.

Eating always seems to be a concern for newly diagnosed families. Talli eats inconsistently. We think that a lot of it has to do with her severe food allergies. She had a g-tube placed when she was 6 months old as she was failure to thrive. Her growth only improved once all of her allergens were removed from her diet and her esophagus was allowed to heal (she had erosive esophagitis). She eats a lot more when she can have whatever she is craving at the time (right now it is Taco Bell) and when she is around her friends. She consistently eats about a third of her calories by mouth. She is learning to weigh her food and typically does her own feedings at school, if they are needed, with her nurse supervising her. She is also learning to read labels for her allergens and protein content.

Brittany Smith brittanydague@gmail.com

Biomarkers for Neurological Injury in PA

Seeking cooperative children and adults for MRI study in Washington DC to see impact of PA on brain markers. Subjects must be able to travel to DC and be stable and do MRI without sedation.

Travel costs and incentive paid. Study takes one day and involves advanced MRI and cognitive testing.
Contact Ileana Pacheco ip126@Georgetown.edu or
Dr. Gropman at agropman@CNMC.org

Help Us Find the Cure!

*Name*_____

*Address*_____

City, State, Zip _____

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Please send an acknowledgement to:

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*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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PROPIONIC ACIDEMIA FOUNDATION

Please email paf@pafoundation.com to join a committee.

PAF VOLUNTEERS AND BOARD MEMBERS ARE NEEDED!

PAF@PAFOUNDATION.COM

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