

Propionic Acidemia Foundation

VOLUME 1, ISSUE 22

SPRING 2017

Deerfield High School School Chest 2016 raises \$168,332.25 for PAF!



We feel an incredible debt of gratitude to the amazing students at DHS who worked tirelessly on every last detail to make School Chest a huge success. In addition to fundraising, School Chest spread awareness about PA among the students and to the local community and provided activities for those with disabilities in which some families affected by PA were able to participate.

Some highlights were:

- Largest School Chest Fundraising Year Ever
- Largest Single Fundraiser for PAF
- Increased outreach efforts by educating over 2000 students, faculty, families and the Deerfield community about PA
- 2 newspaper articles about PA, PAF, and DHS
- Benefit Dinner with silent auction in which Dr. Barbara K. Burton spoke about Jordan's Journey and the need for funding rare disorders
- Skate for School Chest brought PA families and other families with disabilities together with the Warrior Buddies, DHS's peer buddy program, for an ice skating evening to honor Jordan Franks (right) (continued on pg. 6)



Talli and Shayna

**2017 PAF Warrior
Wisdom Conference**
for Families, Medical
Professionals, and
Researchers

June 8th-10th, 2017
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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.

IN MEMORY OF CADENCE

Cadence, June 15, 1996 - January 31, 2017



My sweet Cadence was born on Saturday, June 15, 1996 at 12:39 AM. It was a long, hard labor for both of us, and she had a little trouble transitioning. She was sent to WVU Children's hospital in those first hours, for observation, and came back home to me (still recovering from the ce-

sarean that got her here) a few days later. I did not get to hold this precious blue eyed child until she was 4 days old, but once I did, I vowed nothing would separate us again. Two weeks later, PA reared its ugly head and sent Cadence back to WVU for 32 days. These were harrowing times, no one caring for Cady had ever seen anything like it, and it was a very long time before we realized what we were dealing with. I had just graduated with dual degrees from college literally two weeks before Cady was born, and my strength just happened to be in organic chemistry. The learning curve caring for Cady and understanding her disease was steep, but my background allowed me to understand it deeply and assume her care.

Over the years, despite PA, Cadence had a very full and happy life. I refused to let this monster steal a second of our lives and was determined to give Cady everything within my power to ensure a wonderful life for her. She loved to travel, she has literally been from the East Coast to the West Coast of the United States. She had more friends than can be counted. She laughed and played every waking minute of her life. She was so full of joy and laughter. Over the last weekend of January, Cadence's belly began to hurt and she had trouble having bowel movements. Ileus was common for Cady and we'd weathered several in the last few years. She was managing Saturday, but something changed Sunday. Her pain became more distracting to her, and she asked to go to the hospital. She always knew when it was time. Once at the local ER, ileus was seen on x-ray and an NG tube was placed

to relieve the pressure on her stomach. She was immediately relieved, and was once again smiling and happy. She was loaded into an ambulance for transport to WVU for observation. That was the last time I'd see her smile.

Two hours later, upon arrival at WVU, Cadence crashed and had to be rushed to emergency surgery. Surgeons discovered not an ileus, but a volvulus, where the bowel twists and cuts off the blood supply to the gut. Without blood, her gut began to die off, and surgeons removed 8-10 inches of bowel trying to save her. Over the next few hours, it was touch and go, but Cadence was so strong, she actually was managing to get better. In the end, however, the sepsis proved to be too much, she never regained her blood pressure, and that led to further death of her gut. We were given the option to stop these extraordinary

measures and give her a peaceful passing. As I stood at the foot of Cady's bed, rubbing her feet (because that's all she ever wanted when she didn't feel good), the rest of the world slipped away and it was once again just she and I. I closed my eyes as I held her feet and listened to her heart tell my heart, "No, Mommy. I just want to play. I don't want to do this anymore." And so, as the last act as her advocate, as her protector and mother, I gave her what she wanted, and returned her to her Creator. We stopped the drips and in just a few minutes, her heart stopped. At 3:35 pm on Tuesday, January 31, 2017, my

sweet Cadence threw her arms around her Creator's neck and laughed in utter joy. No more g-tubes, no more nausea, no more belly aches, no more fatigue. Cadence now only knows joy and my heart sings to know this. I never did anything so grand as to deserve the gift that God bestowed on me by giving her to me. She was a blessing from beginning to end, and continues to be so.

See *Cady's Story* in *Fall 2016's issue*.



PA Registry

Help move research forward for propionic acidemia. Participate in the Propionic Acidemia International Registry. As of February 1st, there are 49 participants. For more information on joining the registry, or to update your information, go to www.paregistry.org

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.

The Propionic Acidemia Foundation does not recommend nor endorse any particular products, companies, or manufacturers.

CLINICAL AND BASIC INVESTIGATIONS OF PA, NIH PROTOCOL



DEPARTMENT OF HEALTH AND HUMAN SERVICES

Public Health Service



NHGRI/NIH
 10 Center Drive
 Building 10 CRC, Room 7N248, MS 1646
 Bethesda, MD 20892
 Phone: 301-827-6529
 Fax: 301-480-1721
 Email: pastudy@nih.gov

Summary

This study will evaluate patients with propionic acidemia (PA) to learn more about the genetic and biochemical causes of this inherited metabolic disorder and the medical complications associated with it. People with PA may have problems with learning and development, and may also have heart, gut, kidney, and other problems. They can become seriously ill, sometimes with little warning. There is no cure for PA, but special diets and transplantation are used for treatment.

Participants are admitted to the NIH Clinical Center for 4-5 days each year for 5-15 years for the following tests and procedures:

- Medical history, physical examination, eye examination;
- Consultations from health professionals and doctors who are experts in the nervous system, digestive tract, endocrine or growth glands, kidney, blood cells and immune system, nutrition, physical medicine and rehabilitation or other areas of medicine as needed;
- 24-hour urine collection to examine for electrolytes, organic acids, amino acids, sugar, and proteins for measuring kidney function;
- Blood tests to assess liver and thyroid function, blood counts and blood chemistries, and for genetic tests and basic research studies;
- Metabolic cart study to measure calories needed at rest (resting energy expenditure);
- Dental and saliva evaluation to determine acid levels and dental health;
- Photographs of the face and body (wearing underwear) to help track growth and appearance;
- Ultrasound of the abdomen;
- Electrocardiogram and ultrasound of the heart;
- Hand x-ray to determine bone age;
- Dual energy x-ray absorptiometry (DEXA) scan to evaluate bone density;
- Electroencephalogram (EEG) to study the electrical activity of brain;
- Brain magnetic resonance imaging (MRI) and magnetic resonance spectroscopy (MRS) if it has not been performed in the last year and there are neurological concerns;
- Skin biopsy for cell culture (cells to grow in the laboratory for future testing) if not yet performed or unavailable. Not all participants will need this testing;

- A stool sample to study how gut microbes affect propionic acidemia;
- Some patients may participate in a stable (non-radioactive) isotope study to help understand how body processes propionic acid;
- Other medical tests or procedures recommended by consulting doctors, if medically indicated.

Any patient who becomes seriously ill during the evaluation may be cared for at the NIH or transferred to another hospital if it is deemed advisable. Care provided to you (your child) while at the NIH cannot replace the medical care you (your child) need from local doctors.

Criteria for study participation

Inclusion criteria

- Patients 2 years of age and older are eligible to enroll in this study.
- Patients with biochemical, molecular or enzymatic evidence of propionic acidemia of any gender and ethnicity are eligible to enroll in this protocol.
- Patients with suspected genetic but unknown types of propionic acidemia may also be invited to participate.

Exclusion criteria

- Patients will be excluded if they cannot travel to the NIH because of their medical condition.
- Pregnant patients are eligible to participate in the study. However, they may not take part in some tests, for example stable isotope studies.
- The principal investigator may decline to enroll a patient for other reasons. Other criteria that may lead to exclusion include, for example, residing in a hospital, sub-optimal metabolic control as determined by Dr. Venditti's review of the laboratory data, any patient who requires dialysis once or more in a week and weighs less than 40 kg, any patient who is being treated for an intercurrent infection with antibiotics or has evidence of an acute infection, and any patient who does not have a regular/local metabolic, genetic or endocrine physician and/or a family physician, pediatrician, or internist.

Contact information

Susan Ferry, RN, BSN
 Research Nurse Specialist
 NHGRI/NIH
 10 Center Drive
 Building 10 CRC, Room 7N248, MS 1646
 Bethesda, MD 20892

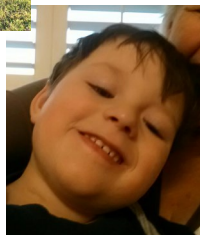
Phone: 301-827-6529
 Cell: 240-381-1717
 Fax: 301-480-1721
 Email: pastudy@nih.gov

GRANT & SEBASTIAN



Our story with PA started out the same as many of yours. Our sons were born looking perfectly healthy, besides being born six weeks early and being twins. Everyone that saw them thought they looked wonder-

ful and would go home from the NICU rather quickly. However, that all changed on the sixth day of their life. I got a call from the doctor in the NICU that Grant was struggling and had to be put on a ventilator, but they thought it was just a virus of some sort. By the time I got to the hospital he was completely comatose and there were swarms of people around him. They were trying to explain exactly what they thought had happened, but all I could understand at the time was that he was very sick and they didn't know what to do. They were running a lot of labs on him and his twin brother Sebastian to try and see if both boys had a metabolic issue of some sort. We were quickly transferred to another hospital that had dealt with these types of conditions before. While I can remember the whole day perfectly now, in the moment everything was a blur and I seemed to be just a spectator as they hooked both boys up to an abundant amount of machines that seemed so humongous in comparison to their little 4lb bodies. They were too small for traditional dialysis, so they attempted to come up with a plan. Fortunately there was actually a visiting geneticist who was interviewing for a position at



the hospital that day, who mentioned he had heard of combining ECMO and dialysis to help patients who were very small. This is what was decided as the best course of treatment for our sons. I will never forget standing over my son as they hooked up IV's and poked and prodded him, all without him making a single, solitary sound. The ECMO and dialysis combination worked, even better than the doctors had anticipated and it seemed that both boys were on their way from catabolic to anabolic. Over the course of the next few weeks, we learned all about PA and the life that would now be our new normal. It was quite the operation at our house to get them fed around the clock, once they came home, thankfully we had and continue to have amazing support from family. The boys have had many hospitalizations since they have come home and have definitely given me lots of new gray hairs, but they have come

so far. While our normal is being hospitalized with one or both of them at least once a month, they continue to grow and show us just how determined they are. They both started preschool this year and are riding the bus everyday to school. Anyone they come into contact with quickly becomes a new friend. It is so amazing to see how positively they affect the people that they come into contact with. It's as if their personalities and happiness are contagious.

Mom of Grant & Sebastian,
3 year old PA Superheroes



2017 PAF WARRIOR WISDOM CONFERENCE

2017 PAF Warrior Wisdom Conference
for Families, Medical Professionals, and Researchers
June 8th-10th, 2017

Deerfield, IL

Hyatt Regency Deerfield: 1750 Lake Cook Road, Deerfield, IL

Get updates on current research and treatment and plan on next steps to realizing our mission of better treatments and a cure for PA.

- Group rate: \$99.00+ tax includes breakfast for up to 4 people.
- Online <https://aws.passkey.com/go/PropionicAcidemiaFoundation>
- You may also call the Hyatt reservation hotline at (888) 421-1442 and they may be able to further assist you as well.
- Make your reservations by May 9th to get the group rate.
- The Hyatt Regency Deerfield is located in the North Suburbs of Chicago. You may want to extend your stay for shopping, Six Flags Amusement park, Chicago Botanic Gardens or Navy Pier

For more info contact PAF: paf@pafoundation.com or 877-720-2192. (Registration on page 5)



Warrior Wisdom Conference Registration Form

PAF 2017 June 8th-10th, 2017 Hyatt Regency Deerfield, Deerfield, IL

CONFERENCE REGISTRATION DEADLINE MAY 9: Please complete and return no later than May 9, 2017. Your prompt registration will assist us in planning this event. If you provide an e-mail address, a registration confirmation will be e-mailed to you.

Name: _____ PA? Yes No

Address: _____ City: _____ St: _____ Zip: _____

Email Address: _____ Phone: _____

Will you be staying at the Hyatt Regency Deerfield? Yes No

Arrival date: _____ Departure date: _____

Names of children attending with you:

Child's Name: _____ Age: _____ PA? Yes No

Low protein meal needed? Yes No Allergies? Yes No If yes, please specify _____

Child's Name: _____ Age: _____ PA? Yes No

Low protein meal needed? Yes No Allergies? Yes No If yes, please specify _____

Child's Name: _____ Age: _____ PA? Yes No

Low protein meal needed? Yes No Allergies? Yes No If yes, please specify _____

Child's Name: _____ Age: _____ PA? Yes No

Low protein meal needed? Yes No Allergies? Yes No If yes, please specify _____

Others attending:

Name: _____ Relationship: _____

Name: _____ Relationship: _____

Name: _____ Relationship: _____

Name: _____ Relationship: _____

Is this your first PAF event? Yes No

Are you a professional or vendor attendee? Yes No

Please note any special needs: _____

Registration Fees (registration includes lunch on Friday and Saturday):

There is no charge for affected individuals or their immediate family.

Additional family and friends: \$75.00/person.

Researchers and medical providers: \$75.00/person (contact pafboard@gmail.com)

Industry representatives: \$125.00/person

TOTAL ATTENDING: _____ TOTAL Registration Fee Enclosed: \$ _____

Registration fees can be paid through Paypal to paf@pafoundation.com or mail to:

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

**For sponsorship
information e-mail
pafboard@gmail.com**

**Thank you and we look
forward to seeing you at
the conference.**

PAF EVENT & FUNDRAISING SPOTLIGHT

PAST EVENTS & CAMPAIGNS:

- **Annual Giving Campaign** - \$14,675 and counting
- **Deerfield HS School Chest** - \$168,332 and counting
- **Tara Gerlach-Columbus Half Marathon 2016** - \$470
- **Ohio Scrapbook Crop** - \$2162
- **Branch Family Superbowl Party** - \$800

UPCOMING/ONGOING EVENTS

- Sep 30- 12th Annual Tailgate Party & Corn Hole Tourney

CORPORATE MATCHING GIFTS AND VOLUNTEER HOURS DONATIONS:

- **Corporate Matching Gifts:** This may enable you to double your donation. Check with Human Resources to see if your employer matches. It makes a big difference.
- **Volunteer Hours:** Some companies have a volunteer program and will donate based on your volunteer hours. PAF is always looking for volunteers. Please check with Human Resources to see if they have a program.

ONLINE TOTALS RAISED TO DATE:

- Igive: \$4,631.67 (Aug. 2016– Jan. 2017, \$106.87)
- Goodsearch: \$ 1,411.03
- AmazonSmile: \$180.17 as of Nov. 2016

DEDICATED GIFTS FROM INDIVIDUALS:

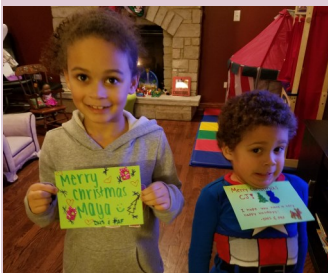
Among the many contributions received, the following is a list of some that were dedicated to those who have inspired the giver.

In Honor Of: Baylor Leone, Peter Savvas, Laura Lemire, Kristin Boecker, Gwen Mouat, Zacharia Matz, Gabrielle Millett, Anna Cardone, Leah Masten, Allison Ellis, Maren Stecken, Lucy Harding, Talli Smith, Dylan Jaehnke, Benjamin Sweetman, Brett, Brayden and Morgan Young, Brandon Napiwocki, Carson Alfano, Trent McKinley, Judy and Steve Lenert, Teegan Sakaguchi, Cohen Waid, Chase Workman, Kate Lowry, Carolyn Schlein, Nila Branch

In Memory Of: Cadence Pierce, Kerrie Fessler, Angelica Stageman, Vincent Franze, Jordan Franks, Bobby Joe Utley, Stella Li, Sharon Esses, Alexa Faith Cardone, Alice and John Dawe, Connor McKillop, Sean Patrick Callahan and Courtney Leigh Callahan, Nicholas Phillips

If you have anyone you would like to have us add to our campaign, please e-mail paf@pafoundation.com

WARRIOR BIRTHDAY CLUB & SCHOOL CHEST (CONT.)



Rob and I were completely blown away and amazed at everything the students at Deerfield High School did to raise the funds for the Propionic Acidemia Foundation.

Rob and I work with students on a daily basis, and were still in awe at the commitment and dedication these kids gave to help others. Maya and Christian loved getting their cards in the mail. As soon as they saw their names on the outside envelope they started jumping up and down. They loved the personalized message and cute stickers. We will forever be grateful to these students! -Heather

Here is the photo of Nila with the Christmas Cards :) she absolutely loved getting mail from a new friend!!! Thanks so much! -Tina



(continued from cover)

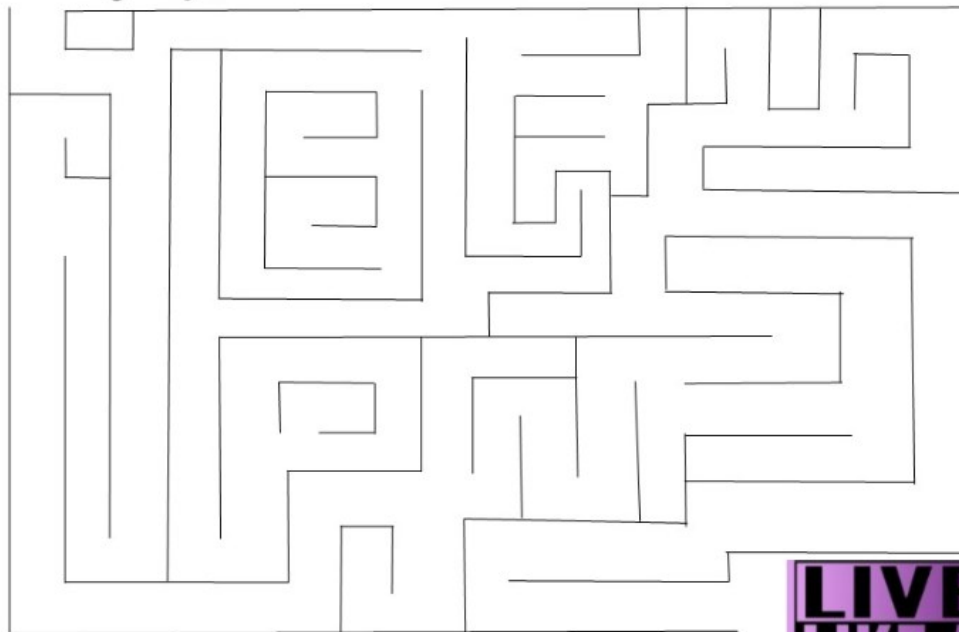
- Jordan's Day was hosted at the school where students made over 100 birthday and holiday cards for children with PA and their siblings (left)
- The sophomore class hosted an evening game night with board games, pizza and candy galore
- 300+ walkers/runners came out for 5K Run for Change color run/walk (above)
- Jordan's Playground was a new event for children to come and play on a cold winter day
- Multiple dine-out nights where local restaurants donated a portion of their sales and over 60 in school food sales
- Fitness enthusiasts were able to donate and attend various classes including WERQ it for School Chest, yoga, cardio kickboxing, spin and barre classes
- Fashion for Compassion had local businesses give a portion of sales
- Deerfield Voice had amazing student and staff performances and was honored to have celebrity guest Isaiah Grass as a judge and performer



DHS 5K: Jill, Brittany and Heather

We are incredibly grateful for the dedication and commitment of DHS; PAF and our families will forever be changed. Thank you again DHS from the bottom of our hearts. The money raised will go toward research and the Warrior Wisdom Conference on June 8th -10th, 2017.

Make your way to the PAF Warrior Wisdom Conference



Clipart by Norma Brook provided by Creative Commons license



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.

We want to hear from you! Have a PA story to tell, event to promote or news? Fall newsletter submissions due by July 31, 2017!

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 _____

- By providing your e-mail address you are opting in to receiving e-mails from the Propionic Acidemia Foundation. We will not share your information with those outside of the foundation.
- 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



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**PAF volunteers and
board members are
needed!**

paf@pafoundation.com



Join and follow
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