



# PROPIONIC ACIDEMIA FOUNDATION

SPRING 2025

SEARCHING FOR A CURE HOPE FOR OUR CHILDREN

## Advancing Research and Care: Propionic Acidemia Conference

July 11-12, 2025  
Kidron Community Center  
4434 Kidron Rd  
Dalton, OH

### PROGRAM

#### Friday, July 11 - 4:00-7:30 PM

4:00-7:30 PM Registration, light dinner, parent panel and discussion, visit exhibitors

#### Saturday, July 12 - 8:30 AM-4:00 PM

8:30-9:00 AM Registration / Exhibits Open

9:00-9:15 AM Welcome/Introductions

9:15-10:00 AM PA Overview/Research Charles P. Venditti MD, PhD, NHGRI, NIH

10:00-10:30 AM Break - Meet other families and visit exhibitors

10:30-11:15 AM Metabolic insights into PA: pathological mechanisms and emerging therapeutic strategies, Guofang Zhang, PhD, Duke University

11:15-11:45 PM TBD

11:45-1:00 PM Lunch

1:00-1:15 PM Cardiovascular Pathology in PA Across Zygosity Type, Andres Hollenbeck, Cardiology Care for Children

1:15-1:30 PM Electrophysiological Abnormalities in PA: ECG Analysis Across Genotypes, Carla Leal, BCN, Medtech group Physense

1:30-1:45 PM Machine Learning-Based Electrocardiographic Analysis to Characterize the Cardiac Phenotype in PA, Carla Leal, BCN, Medtech group Physense

1:45-2:30 PM Developing Medicines to Treat Liver Stop Codon Disease, Nerissa C. Kreher, MD, CMO, alltrna

2:30-3:00 PM Break - Meet other families and visit exhibitors

(cont. PAGE 2)

### PA Registry

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

For more information on joining the registry, or to update your information, go to [www.paregistry.org](http://www.paregistry.org)

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## ADVANCING RESEARCH AND CARE PROGRAM (CONT. PAGE 1)

3:00-3:45 PM mRNA-3927 for PA: Aggregate Interim Results of Ph 1/2 and Long-Term Extension Trials, Rosa Real, MD, Moderna

3:45-4:00 PM Wrap up

\*Program subject to change

**A limited number of no-cost Echocardiograms and EKGs for the patients and families with PA will be available. To schedule, contact Daniela at 717-925-8300.**

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## PA HOPE IN THE HEARTLAND FAMILY CONFERENCE SUMMARY

On October 19th, 2024 the Propionic Acidemia Foundation and the Community Health Clinic cohosted the PA Hope in the Heartland Family Conference. We are thankful to our generous Gold Sponsor Moderna and Silver Sponsor Nutricia.

The conference was held at The Community Health Clinic in Shipshewana, IN.

Speakers included Dr. Zineb Ammous, Dr. Joshua Meisner, Dr. Charles P. Venditti, Dr. Kyle Soltys, Dr. Anthony Zaki, and Elaine Wakefield. Thanks to technology this was our first hybrid meeting and was attended in person by 122 people (25 affected families) and on Zoom with 31 people. Zoom attendees had the opportunity to participate and ask questions and join the discussion. Attendees were from 14 states and 5 countries.

Recordings of the presentations are available at [youtube.com/@propionicacidemiafoundation2372](https://youtube.com/@propionicacidemiafoundation2372)



## ELIZABETH & MORGANE

Our story starts out like so many others... fast-forward to six-year-old Elizabeth. By now, Elizabeth has had more hospital stays than most children have in their entire lives, more than I would like to recall. She gained a little sister not long after she turned two years old.

In October 2022, six-year-old Elizabeth had been very sick and wasn't going to get better unless she had IV fluids and stronger medicines. When we got to the ED(emergency department) she was expedited through the lobby to a room to start on her metabolic protocol. Elizabeth's ammonia was higher than normal, which is always concerning. She was sent to the floor. From the viral panel ran, they found that Elizabeth had a rhinovirus. We weren't on the floor for very long before we were moved to the PICU.

Once they assessed Elizabeth, it was decided that she needed to start CRRT(dialysis). It wasn't long before the doctors said the CRRT wasn't enough and we had to get her on ECMO (extracorporeal membrane oxygenation). They said that if they had waiting only a few more minutes, they would have had to start CPR.

Elizabeth was on ECMO/CRRT for 11 days. Her eyesight took a hit when she was working so hard to breathe before moving to the lifesaving machines. Because of this lack of oxygen, her optic nerves took a hit, and left her legally blind.

After her ECMO stay, we were in and out of the hospital for several months and always seemed to end up in the PICU. During one of these stays, the team said it was time to have the liver transplant conversation. Never in my wildest imagination, did I think we would have had that conversation so soon.

The liver transplant evaluation, took place over several days while Elizabeth was inpatient. This entailed numerous lab draws and meeting with different parts of the team. It was determined that Elizabeth was a good candidate for a liver transplant. It took less than 20 days, before we got 'the call'. After arriving, every-thing went quickly and smoothly. The team was well prepared.



Morgane & Elizabeth (left), Morgane, Elizabeth, Quinn & Isaac (right top from Kay Dodds Photography), Elizabeth & Morgane (right bottom)



Being metabolic, they had each step written in detail. Many of the transplants are done in the night, due to the OR being occupied during the day. Elizabeth received a new liver, thanks to the generous/incredibly unselfish gift that has drastically changed her life for the better.

It took 6 weeks to get back home from the transplant. Elizabeth has had a few rejection scares. Rejection is a scary word, but they have been able to work things out to where the rejection subsides.

It's been nearly 2 years after the transplant, Elizabeth's liver labs are checked every 2 months now.

The liver team would ask about Morgane at most liver appointments. I said we needed to get the first year of Elizabeth's transplant under our belt before we were ready to talk about Morgane getting one.

Morgane completed her liver transplant evaluation the summer of 2024. She was listed at the end of 2024. The liver transplant program became inactive in December 2024. They are in search of another surgeon to join the team before they are active again. When they are active again, there will be labs drawn again and weight/height taken to be reactivated on the transplant list. So until that happens, we will work to keep Morgane healthy and be prepared for when we get the call for her.



## NAYELI

This is Nayeli. She was born on July 3rd, 2024. We had the most amazing pregnancy and delivery you could imagine. My husband, Luis, and I were so excited to find out we were having a baby girl. We immediately started picking out names and started planning our daughters nursery. We went to so many stores and got loads of pink items, dresses, Stuffed animals, etc. We made a promise to always make sure our baby girl had everything she needed and all the love in the world.

The night of delivery went smoothly for the most part. Nayeli's heart rate was beginning to drop with contractions which had us both worried. She was here within an hour. After delivery, we began to see lots of complications with her. Her sugar levels were dangerously low, and her body temperature would not regulate. She stayed with me in Mother baby the first night, and we struggled to feed her. She was not interested in bottle feeding and refused to latch. I couldn't help to feel guilty and thought maybe its my fault, I am doing something wrong. The nurses would assure me that she will be okay and she just needed time.

The next day came around and Nayeli continued to get worse. She was starting to have vomit episodes and still would not regulate her body temperature. Thursday night she was taken upstairs to the NICU where they gave her an NG tube. They started running tests on her to figure out what was wrong.

First we were told she had a virus, then a blood infection, and that hopefully with antibiotics she will get better. I had a gut feeling this was not the case and something else was starting to become terribly wrong with my baby. She was at Edwards Hospital NICU for a week, where they put her on fats/lipids through her IV. After this Nayeli began to deteriorate very quickly. A day after this, she was put on oxygen and would no longer open her eyes or move any part of her body.

It was around 12:33 am when we got a phone call, where the doctor told us her Newborn screening came back flagged for PA, and that she was being



sent to Ann & Robert H. Lurie Children's Hospital of Chicago. Her ammonia level was very high and once she arrived she was placed on Dialysis and was intubated. We were prepared for the worst. My husband and I cried so much during this time and were so terrified that our baby girl would not get to come home.

Nayeli is a warrior. She fought so hard and after 3 very long months of being in the PICU & NICU, she was finally able to come home. Our baby girl is a rock star. With her daily medicine regimen and formula plan she is now doing very well and continues to be stable. She is in physical therapy to help meet her milestones and we couldn't be more happier with her progress. We are thankful for everyone who has supported us and is continuing to support us and other families with PA.



Nayeli, Luis and Mari

## RARE DISEASE DAY ARTWORK SUBMISSIONS

Thank you to Miguel, Olivia, and Ray for your creative submissions. Products still available at [Bonfire.com/store/propionic-acidemia-foundation/](https://bonfire.com/store/propionic-acidemia-foundation/)



## BE PREPARED

### Connect with local utility and emergency services & sign-up for emergency alerts

- Many municipalities will maintain a copy of an Emergency Protocol with the emergency services or paramedics, so that the information is on hand if they are called.
- Contact your local utilities to inform them of medical needs for power for refrigeration and/or feeding pump, so that they can put your household on their priority list in case of the loss of power.
- Sign-up for local government & school alerts, so you are notified about local emergency situations.
- Consider maintaining a landline telephone, as it does not depend upon electricity coming through powerlines. Cell phone towers can become overloaded during emergencies and natural disasters.
- Enable wireless emergency alerts on your cell phone.
- Enable alerts on the weather app on your cell phone.
- Enable emergency alerts on Alexa smart home devices (Google devices do not have this feature).
- Sign-up with Medic Alert. They have multiple plans that include your health profile, but also notification of family members, information about implanted devices like ICDs, and keeping an Emergency Protocol on file. (Medicalert.org)



## CLINICAL RESEARCH STUDIES ON PA – an update

### What is happening in clinical research for PA?

#### ENROLLING PATIENTS

##### **MODERNA: Phase I/II Clinical Trial on mRNA-3927 for Propionic Acidemia**

- On April 3, 2024, Moderna published the first data on the first ever clinical trial with mRNA therapy for PA.

- So far, the trial has included 16 participants, with over 340 doses of the treatment administered.
- No significant Adverse Events have been reported.
- Preliminary findings report that the relative risk for metabolic decompensation events were reduced by 70% compared to patients that did not receive the mRNA.

Most participants are now continuing into the open-label extension study (all participants will receive now the therapy). <https://investors.modernatx.com/news/news-details/2024/Phase-12-Interim-Data-on-Modernas-mRNA-3927-an-Investigational-mRNA-Therapy-for-Propionic-Acidemia-Published-in-Nature/default.aspx>

##### **NIH NATURAL HISTORY STUDY ON PA.** Dr. Venditti et al.

-Dr. Charles Venditti and his group at the National Institute of Health (NIH) continue the Natural History study on PA, started in 2016, to better understand PA and design better treatments for the disease.

Patients as young as 1 month-old spend 5 days at the NIH and undergo a battery of studies, from blood tests to brain imaging, and consultations with numerous specialists, to collect data on PA.

The study is still recruiting new participants as well as doing follow up visits with existing patients.

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#### IN THE PIPELINE

**PaVe-GT pilot project to increase the efficiency of gene therapy** (NCATS, National Human Genome Research Institute, NINDS, Eunice Kennedy Shriver of Child Health and Human Development).

- The PaVe-GT program is testing and optimizing the efficiency of AAV9 vectors as a platform for clinical trials with the aim to expand it to many other diseases.
- The PCCA gene for Propionic Acidemia is one of the 4 different diseases where the platform is being tested with the intention to go into clinical trials in the upcoming years.

**The Bespoke Gene Therapy Consortium (BGTC) chose the PCCB gene for Propionic Acidemia as 1 of 14 candidates for gene therapy clinical trials to advance gene therapy trials for rare genetic disease.**

- The consortium is a public-private partnership involving NIH institutes, FDA, pharmaceutical and biotech companies and non-profit organizations.

<https://fnih.org/news/amp-bgtc-announces-rare-disease-selection-candidates-issues-clinical-trial-rfp/>



## CLARIVATE STUDY ADVERTISEMENT



### Study advertisement

Clarivate (a research consultancy) are looking for

- Adults ( $\geq 18$  years) who are diagnosed with propionic acidemia
- Adolescents (13-17 years) who are diagnosed with propionic acidemia
- Caregivers of children (aged 0-17 years) diagnosed with propionic acidemia

in the US, UK, Germany, Japan and Italy to take part in a 60-minute interview.

The purpose of the interview is to explore your and your child's experience of living with propionic acidemia.

Participation is entirely voluntary, and your personal information would be kept confidential. The interview will either take place online, using Microsoft Teams, or face-to-face. You would be compensated for your time.

If you are interested in taking part, or would like more information, please contact Clarivate at [PAinterviewstudy@clarivate.com](mailto:PAinterviewstudy@clarivate.com).

## REQUEST FOR PROPOSALS

### REQUEST FOR

### RESEARCH PROPOSALS



### PAF: ADVANCING RESEARCH AND IMPROVING LIVES

PAF is a non-profit organization that is committed to advancing research and finding better treatments, and ultimately a cure, for propionic acidemia. Our primary aim is to fund projects which will accelerate new knowledge about PA, promote the discovery of biomarkers and co-morbid conditions, and develop and evaluate therapeutics that can help improve the lives of those affected by PA.

PAF will entertain any proposal with the potential to advance treatments and improve the lives of those with PA

### APPLICANT QUALIFICATIONS:

To be considered, candidates must possess a PhD, MD, or equivalent degree, and currently hold a full-time position at an established academic or research institution, regardless of their current rank (post-doctoral, research scientist, professor, etc.).

**DEADLINE: OCTOBER 1, 2025**



### GRANT SPECIFICATIONS

[www.pafoundation.com](http://www.pafoundation.com)  
[paf@pafoundation.com](mailto:paf@pafoundation.com)  
 877-720-2192

## PAF ACTIVITIES AND FUNDRAISING SPOTLIGHT

### UPCOMING EVENTS

- Low Protein Community Virtual New Parent Cafe-4/27
- Presenting at ASGCT Meeting - New Orleans 5/16
- Presenting at BIO International Convention - Boston, MA 6/17
- PAF Conference, "Advancing Research and Care: Propionic Acidemia Symposium" Dalton, OH 7/11-7/12
- OH Families Fall Fest - TBD
- The TCS New York City Marathon 2025 - 11/2

### PAST EVENTS

- Ohio Families Fall Fest 2024 raised \$8200
- The TCS New York City Marathon Runners 2024 raised \$27,912
- PAF exhibited at PKU Organization of IL and Allied Disorders Low Protein Annual Meeting 11/9/2024
- PKU Organization of Illinois and Allied Disorders New Parent Cafe (page 10)

### MATCHING DONATIONS & VOLUNTEER HOURS

This may enable you to double your donation. Check with Human Resources to see if your employer matches. Some companies have a volunteer program and will donate based on your volunteer hours. PAF is always looking for volunteers.

### INTERNET

Thank you for using iGive, Goodsearch, and Bing and selling on eBay. Every dollar helps.

### DEDICATED GIFTS FROM INDIVIDUALS

- IN HONOR OF: Alexa Faith Cardone, Allison Ellis, Dylan Jaehnke, Laura Lemire, Kate Lowry, Reily and Judson Lenert, Leah Masten, Trent McKinley, Gabrielle Millett, Gwen Mouat, Chase Workman
- IN MEMORY OF: Sean and Courtney Callahan, Alice and John Dawe, Kerrie Fessler, Jordan Franks, Vincent Franz, Christine Malys, R.J. Malys, Elizabeth Malys, Connor McKillop, Michael Messersmith, Brandon Napiwocki, Jean Philips, Nicholas Phillips, Angelica Stageman, Kristin Boecker, Tallina Smith, Teegan Sakaguchi, Kirstyn Tripp, Tyler Weinzerl

### FACEBOOK

Thank you to all of our Facebook Fundraisers and people that donated to their fundraising pages for birthdays, #GivingTuesday or just because: Jill Chertow, Dottie McGee, Karen Eddy-McVey, Cindy Olhoft, Sarah Mullins, Andrea Sherwin, Jenn Simmons

### STOCK DONATIONS

PAF accepts stock donations. Please email [paf@pafoundation.com](mailto:paf@pafoundation.com) with any questions.

## 2ND ANNUAL OHIO FAMILIES FALL FEST NOVEMBER 2, 2024



### NEW BOARD MEMBER!!


.....

The Propionic Acidemia Foundation Board of Directors

WOULD LIKE TO WELCOME...

**Jennifer Simmons**

NEW YORK





## 2025 TCS NEW YORK CITY MARATHON -PARUNNERS 2025



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**November 2, 2025**

**stay tuned for team  
PARUNNERS2025**

for more info email Marisa at [teamPAR4@gmail.com](mailto:teamPAR4@gmail.com)





## OUTREACH-NEW PARENT CAFE

On March 9th, Brittany Smith and Jill Chertow attended the PKU Organization of Illinois and Allied Disorders New Parent Cafe. We were excited to spend time with everyone and thrilled that it was attended by six families affected by propionic acidemia, six with PKU and one with HCU.

It is so important for families to have access to local groups in addition to the global groups. Families had a chance to share their stories, get tips and recipe ideas, and visit with Nutricia and PTC Therapeutics.

Check out the upcoming cohosted Low Protein Community Virtual New Parent Cafe (below) and flock Camps and diet app.



**flok Together: Camp and App with Us!**



flok is advancing research and care for people with Propionic Acidemia. **The flock app** helps you track how you eat, feel, move, and play to offer new insights into our conditions.

Attend our in-person **flok Family Camps** in Oregon and New Hampshire! Our metabolic community members, family, and friends are all welcome for 3 days of fun, learning, and social connection.

Visit [flok.org](https://flok.org) for more information on the flock app and Family Camp registration

## LOW PROTEIN COMMUNITY VIRTUAL NEW PARENT CAFE

**LOW PROTEIN COMMUNITY VIRTUAL NEW PARENT CAFE**

Sunday, April 27, 2025 at 11 am ET

For parents, relatives and caregivers of children ages 0-5 that require a low protein diet. Come network and learn from other families!

Topics will include:

- How to navigate nursing and bottle feeding
- Weaning and introducing solids
- Tube feeding and blenderized diets
- Childcare
- Picky toddlers
- Early intervention programs
- And more!

Registered Dietitian(s) from Chicago leading metabolic centers will also be present!

Logos for PKU Organization of Illinois, MSUD, National Urea Cycle Disorders Foundation, and Orphanet Association are displayed at the bottom.

**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.

**VIISION:** To create a future where Propionic Acidemia can be prevented, and any affected individual can be cured and live a productive life.

## PAF CONFERENCE REGISTRATION FORM - JULY 11-12, 2025

### PERSONAL INFORMATION

Name (first and last) \_\_\_\_\_  
 Address \_\_\_\_\_ City \_\_\_\_\_ State \_\_\_\_\_ Zip Code \_\_\_\_\_  
 Email Address \_\_\_\_\_ Phone \_\_\_\_\_  
 Do you have PA? Yes \_\_\_\_\_ No \_\_\_\_\_ Other diet requirements \_\_\_\_\_

Check all that apply: \_\_\_ Family \_\_\_ Presenter \_\_\_ Exhibitor \_\_\_ Medical Professional \_\_\_ Volunteer  
 Information of others attending with you (add additional page, if necessary)

| NAME (FIRST & LAST) | RELATIONSHIP<br>(SPOUSE, CHILD, ETC) | AGE (IF CHILD) | PA (YES/NO) |
|---------------------|--------------------------------------|----------------|-------------|
|                     |                                      |                |             |
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|                     |                                      |                |             |

Please indicate number of people attending on July 11 dinner: Adults \_\_\_\_\_ Children (3-12) \_\_\_\_\_ Under 3 \_\_\_\_\_  
 July 12 Lunch: Adults \_\_\_\_\_ Children (3-12) \_\_\_\_\_ Under 3 \_\_\_\_\_

Please note any other special needs: \_\_\_\_\_

Dr. Chowdhury's research including FREE Echos/EKG (for the family-affected, carriers, and non-affected), Please indicate how many people: Adults \_\_\_\_\_ Children (3-12) \_\_\_\_\_ Children under 3 \_\_\_\_\_  
 Not Interested: \_\_\_\_\_ (**Limited availability - must contact Daniela - [Dcavet@cardiologylancaster.com](mailto:Dcavet@cardiologylancaster.com) or by phone 717-925-8300**)

I hereby give permission to PAF to use any photographs/videos taken at the 2025 PA Family Conference in which I or members of my family may be a part (for use in, but not limited to, newsletters, PAF websites, social media, and reports). Yes \_\_\_\_\_ No \_\_\_\_\_

In consideration of the acceptance of this registration, I/we the undersigned, assume full responsibility for any injury or accident which may occur while I/we am/are attending the conference events. I/we hereby release and hold harmless the PAF, its officers, directors, staff, volunteers, members, representatives, agents or assigns associated with this event from any and all personal injury, loss or damages.

Signature: \_\_\_\_\_ Date: \_\_\_\_\_

**Registration is due by July 1, 2025.** Please mail completed form to: Propionic Acidemia Foundation, PO Box 151, Deerfield, IL 60015 or register at **[pafoundation.com](http://pafoundation.com)**



Propionic Acidemia Foundation  
P.O. Box 151  
Deerfield, IL. 60015



SEARCHING FOR A CURE  
HOPE FOR OUR CHILDREN

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-  Patient Registry: [paregistry.org](http://paregistry.org)
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