FALL 2023

PROPIONIC ACIDEMIA FOUNDATION

SEARCHING FOR A CURE HOPE FOR OUR CHILDREN

PA Heart to Heart Conference July 2023

Sue Weaver

According to attendance, the 2023 Heart to Heart PA conference was a success. It all began when Steve and I discussed hosting a gathering for families and individuals affected with Propionic Acidemia. How could we make the day educational in a casual, unintimidating atmosphere? What if we could ask some doctors to share their expertise on our comprehension level? Could we offer CPR classes in a simple, straightforward way so folks would dare to take the classes?

We soon decided we couldn't host such a large event at our home. As we considered venue options, Jill Chertow from PAF called to subscribe to our Amish PA Newsletter. We chatted on the phone for a few minutes, and when I mentioned hosting an educational family day, Jill was instantly interested. "What if we would co-host the event?" Jill asked. "Would that be something you'd be interested in?"

Was it ever! During the next year, Jill and I exchanged emails, and spent many hours on the phone discussing what would be most helpful for families. As time went on, and more plans were made, Jill suggested dividing the events into two days. "If families would consider coming for both days, I think we would have more satisfaction that way."

Two days it was. On Friday morning, July 21, we transformed the Rec Center at Kidron Community Park, Ohio into a Propionic Acidemia world. Dr. Devyani Chowdhury and her cardiac team set up three makeshift rooms to offer free echocardiograms, ECGs, and genetic testing. Bart Bijnens from Barcelona, Spain accompanied Dr. Chowdhury's team to share his knowledge.

Families spent the afternoon and evening participating in CPR training provided by EMS personnel from Kidron Volunteer Fire Department. Yoder Veal brought their food truck and cooked supper for us, and everyone brought a side dish, dessert, or snack to share. There was plenty of time to chat with other families and the doctors.

We set up tables along the north wall for our sponsors, CoA Therapeutics, Moderna, HemoShear Therapeutics, Nutricia North America, and Zoia Pharma LLC. The sponsors shared information on their ongoing PA studies, free low protein cookbooks, low-pro foods, and much more. PAF handed out Care Notebooks and Emergency Preparedness Handbooks for PA individuals.

PA Registry

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

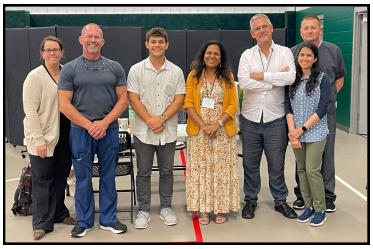
For more information on joining the registry, or to update your information, go to www.paregistry.org

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.

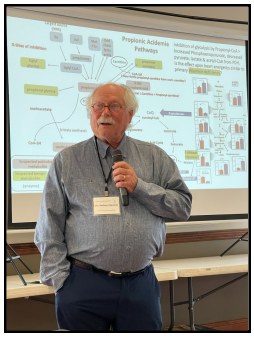
VISION: To create a future where Propionic Acidemia can be prevented and any affected individual can be cured and live a productive life.

HEART TO HEART CONFERENCE SUMMARY - CONT.



Dr. Chowdhury, Bart Bijnens and the Cardiac Team (left)

Dr. Morton and the PA Pathway (right)



The event continued on Saturday, until late afternoon. Dr. Olivia Wenger began the day with the topic, What is PA? She talked about the importance of a low protein diet and shared some excellent recipes for families to try. Bart Bijnens and Dr. Chowdhury talked about their research on the PA heart, and their opinions about treatment. Dr. Holmes Morton's topic delved deeper into gene exomes and bio chemistries of Propionic Acidemia, and the thoughtful therapy approach.

Dr. John Clark and Dr. Brandon Smith, both from Akron Children's Heart Center, shared their fields of expertise. Dr. Clark covered the electrical part of the heart, arrhythmias, and the significance of beta blockers and AEDs. Dr. Smith has been studying PA and cardiomyopathy for years, and shared what he has learned.

The weather was perfect so the children could enjoy the large playground outdoors. Byler's Barbecue provided a haystack meal for lunch, and Red Tomato Market made subs for a grab-n-go supper. And at the end of the day, when Steve and I were beyond exhausted, many hands chipped in to help us clean up.

An astounding three hundred guests attended the event, all eager to glean from the knowledge of experts and communicate with like-minded families. My heart warmed with encouragement and inspiration as I connected with many old and new found friends and conversed with the docs. I didn't know about the rest of the guests, but I felt ready to continue the good fight.

In retrospect, I know all of this wouldn't have been possible without PAF, all the sponsors, medical professionals, and many volunteers who pitched in to help with the set-up and tear-down. On behalf of our Propionic Acidemia community, we offer a humble, but heartfelt thank you!

We will never forget.

LUKAS' AND JOSIAH'S STORIES

"Your baby has Propionic Acidemia."

Our baby has what? Steve and I looked at each other in bewilderment, trying to make sense of what the doctor was saying. It sounded like gibberish.

It was December 2012, and we were sitting in Dr. Kurzynski's office at Akron Children's Hospital with our four-month-old son. Lukas had been born in September at thirty-two weeks gestational, weighing 4 lb. 2 oz. We had spent about a month at the hospital before taking our wee son home. Armed with a monitor to alert us of any bradycardias, and doctor's orders to stay away from sick people, we felt prepared to take care of him.

Until we received the message from his pediatrician. "Lukas' state screening flagged a problem. You need to see a geneticist as soon as possible." I wasn't too concerned about it—we'd been through the same thing with our oldest son, Josiah. But this time, the doctor informed me that I should be worried, and that we need to see a geneticist in Akron.

After Lukas was diagnosed, Dr. Kuczynski dropped another bomb. "What about your other children?"

We knew the results before they came back. Our oldest son, Josiah (age 6), who had struggled all his life was affected; our middle son wasn't. We were right. We did more blood tests, saw a dietician, then a guidance counselor, then the geneticist. He wanted to do more tests, so we went back to the lab again. The next day, Dr. Kuczynski called me. "Your oldest son has dramatic changes in the electrical part of his heart. We need to get him in with a cardiologist asap."

Dr. Clark, the cardiologist, was a wonderful balm for my heart the next day. He took the time to answer all my questions, to educate me, and reassure me that there was medicine to help. Josiah's OTC interval was over 500, so we went home with a Holter monitor. Dr. Clark advised us to get an AED and make sure we knew how to use it.

Those first months were difficult.

LUKAS AND JOSIAH CONT.

Josiah, age 17
Lukas, age 11

We were clueless and confused. Anxious to know more, I drilled the doctors at Akron. They didn't seem to know much about the PCCB variant of Propionic Acidemia other than it was a milder form than the PCCA gene.

One evening, another family with affected children came to visit us. They were our lifesavers. They took the time to explain and make suggestions to help us. They shared their experiences, and told us about Dr. Morton and the Clinic for Special Children in Lancaster, PA.

We stayed home that winter. The doctors warned us that RSV season and a preemie baby with PA aren't a good combination. Lukas was eight months old when he began seizing. The first seizure didn't last long, but he was unresponsive for a while afterwards. We called 911, and an ambulance hurried us to Akron Children's Hospital. Almost as soon as we were in our room, Lukas had another seizure. The nurse grabbed him and ran down the hall to the trauma room, where about ten doctors and nurses surrounded him. A counselor was soon there to ease Steve and me into the hallway so they would have more room to work.

We were admitted to a room on the seventh floor. Lukas didn't sleep well that night, and at four a.m. he had another seizure. And then another one later that morning. An EEG showed no seizure activity, and the doctors were puzzled. Was it PA? Or was it just febrile seizures? Lukas continued to have cluster seizures whenever he was sick or had a temperature until he was almost three years old.

In early spring of 2013, we made the trip to Strasburg, Pennsylvania to see Dr. Morton. That's when we began understanding PA. We spent four hours in the office that day, and Dr. Morton answered all our questions.

He was still doing his study on Pro-Citric, so we went home with several cases of that, and all kinds of other supplements he wanted us to try. We were a little overwhelmed, but so thankful we finally had help. Pro-Citric made a huge difference in our boys' lives. Josiah's muscle tone and his overall health improved.

Josiah had a nasty bike accident a year ago. He was alone when the accident happened, but by all indications he was unconscious before he crashed and had suffered an arrhythmia. After several major discussions with his doctors, he had surgery to implant an ICD.

Lukas had surgery in August to implant a Medtronic Loop Recorder after he collapsed a few times and couldn't remember what happened. The device will monitor Lukas' heart rhythm and send electronic links to his cardiologist every day so they can see what is going on. A relay monitor sits on Lukas' bedside table, and he wears a clicker on a lanyard around his neck that he must press if he feels dizzy or has any weird symptoms. If he collapses before he can click, it's up to us to push that button. The relay monitor sends links automatically, but if the clicker button is activated, it will record additional information for the doctor to see. Hopefully Lukas will be able to wait several years before he needs a pacemaker/defibrillator combo like Josiah has.

Today our boys are cared for by Dr. Morton and Dr. Wang from the DDC Clinic in Middlefield, Ohio. They both have enlarged hearts, and abnormal ECGs— prolonged QTC intervals and inverted t-waves. Dr. John Clark and Dr. Brandon Smith and the team at Akron Heart Center manage their heart care.

Managing PA has become a lot easier over the years, so to all the new families— take heart! Our boys take responsibility for their medicines, formula, and diet, leaving the specifics to Mom. It's hard sometimes to let them go, to refrain from keeping them inside a bubble close to us. But that's not living— so we do all we can and try to leave the rest at God's feet. We are blessed to have a PA community... friends to share the diagnosis with us and doctors who care.

ACE RIGHT-HANDER (REUBEN'S STORY)

The protocol would be painful, a daily schedule of intrusions to be performed, opportunities to probe and poke. "procedures" they were called. I gave my written permission and he became theirs.

We stayed on Rankin, the Clinical Research Unit, two hundred miles from home. Our room was typical hospital décor with a wall-covering of institutional pink. Our one window framed an unsightly view of the central heating plant with its ugly brick smoke stack invading the sky. Off in the distance, the bell tower of Duke Chapel rose through the trees in Gothic grandeur.

During our two week stay, a barrage of medical students, nutritionists, neurologists, psychologists and metabolic specialists found their way to our room. One doctor will be remembered for his grandiose entrance, like a king arriving for his coronation. Another doctor, the Chief of Pediatric Genetics and Metabolism, was as brilliant *as* he was kind. I listened intently, but did not comprehend, his explanation of the "efficiency of certain monosaccharides in providing key metabolic intermediaries to regulate catabolism." My feelings of inadequacy were only intensified by his level of expertise.

The nurses became my soul mates as we complied with orders left behind by men retreating to their research. Making 24-hour urine collections for seven consecutive days from a child still in diapers was a challenge. Inserting a nasal gastrostomy tube into my screaming, kicking baby left me shaking. I felt like a demon. I felt what a nurse must feel at times. I felt strength. I could hurt my child to help him and I would have to do it again.

Our time spent at Duke was outside the mainstream of life with no place to be and no one to see. It passed slowly but it was beneficial to me in unsuspecting ways. It gave me time to be with my "chubby little person", to know him better and to like him more. It also gave me the opportunity to meet the warrior within me, an elusive fighter who only shows herself when battlefield conditions prevail.

Now eight years old, mentally handicapped, hearing impaired and almost exclusively tube-fed, these labels tell little about my son. His metabolic disorder is manageable. Life-threatening crisis no longer occur. The sound of his laughter cackles through our home and is intermixed with the cries of a child who does not understand.

Reuben, age 39

Our son's disorder was the happenstance of heredity, the wrong gene from both parents. He also inherited clear blue eyes, thick brown hair and a passion for the game of baseball. Both his father and grand-father threw balls and strikes for professional teams. Like them, he loves to wear the hat, swing the bat and to throw the ball, most of all. Unlike them, his game will be played on a different diamond. No one will care about the speed or accuracy of his pitch, no one will count the balls and strikes. As my son becomes a man I know his differences will become more pronounced. I can only hope his way of standing on the mound and facing each batter and his wind-up and delivery of the pitch, will be enough to please the hometown fans.

Ace Right-hander was written in 1992 for a creative writing class I was taking at the time.

Reuben will be 40 on his next birthday so it's probably time for an update. He's been very healthy since March 2020. Reuben attends a day program four days a week and a half-day program at our church, once a week.

He loves all things baseball and NFL football. Over the course of many Christmas's and birthdays he's accumulated almost all the team jerseys and hats. We know he's upset about something, which rarely happens, when he throws his hat. Reuben plays on a special needs baseball and basketball team and he loves blue-grass music in all its forms.

To live with Reuben is to be greeted in the morning with "Bless you Mom". His laughter can come for any reason or no reason at all and is as infectious as the common cold. He loves going to church and out to a restaurant, even though he probably won't eat anything, he just loves being around people. Reuben has been assigned the position of is Happiness Co-Ordinator at his sisters' Early Intervention company and is the primary reason she chose to work with children with special needs. (Cont. PAGE 6)

PAF AWARDS $50,000 CONTINUATION GRANT

PROJECT: Elucidation of cardiac electrophysiological alterations in propionic acidemia: Towards the identification of targets for therapeutics. PI: Eva Richard, PhD, Centro de Biología Molecular "Severo Ochoa" and Eva Delpón, Universidad Complutense de Madrid

How the use of induced pluripotent stem cells can help us to model a disease such as propionic acidemia?

For many years our group has been investigating propionic acidemia (PA) disease, contributing to seminal discoveries such as the structure of the PCCA and PCCB genes, genetic epidemiology of the disease in different populations, the functional analysis of mutations in prokaryotic and eukaryotic systems, investigation of the genotype-phenotype correlations, identification of miRNAs as potential biomarkers and the characterization of the cardiac phenotype of the hypomorphic PA mice. Although the PA mouse model has been used to study some aspects related to the cardiac phenotype, differences in heart structure, heart rate and other parameters make it unsuitable for more in-depth studies of the human cardiac alterations related to PA.

Thanks to 3 years of funding from the Propionic Acidemia Foundation (PAF) we have been able to take a step further taking advantage of the amazing potential offered by the use of induced pluripotent stem cells (iPSCs). These cells provide a very attractive tool for researchers as they can differentiate into any cell type. Thus, from PA patients' fibroblasts we have been able to reprogram these somatic cells and obtain patient-specific stem cells. We have differentiated these cells into cardiomyocytes, thus achieving a disease model with a human context. These cardiomyocytes are invaluable because they allow us to study the pathophysiology of the disease and discover new therapeutic targets.

In our previous project (PAF-107) we were able to obtain cardiomyocytes from two patients with defects in the PCCA and PCCB genes. These models allowed us to study the molecular mechanisms that may be contributing to the cardiac phenotype of the patients. These results have been published in two research articles: Alonso-Barroso E. et al (2021).

"Cardiomyocytes derived from induced pluripotent stem cells as a disease model for propionic acidemia". Int J Mol Sci 22: 1161-1175. Álvarez M. et al. (2023). "Dysregulated cell homeostasis and miRNAs in human iPSC-derived cardiomyocytes from a propionic acidemia patient with cardiomyopathy". Int J Mol Sci 24, 2182. Based on the results obtained, we wanted to make further progress and set out to identify the electrophysiological mechanism responsible for the cardiac complications in these PA patients.

For that goal, we contacted the research group led by Professors Eva Delpón and Ricardo Caballero at Universidad Complutense of Madrid who are world-renowned experts in "Cardiac Cellular Electrophysiology" area and, specifically, on the pharmacological and physiological modulation of human cardiac ion channels. When we proposed this work to them, they found it very attractive with a good translational objective since it represents a great opportunity for PA patients to benefit from our scientific knowledge. And this is what we have been working on during this first year of the project (PAF-113) funded by PAF.

Initially, obtaining mature cardiomyocytes was a challenge because it is difficult to obtain this type of functional cells allowing to record action potentials, but after much effort we succeeded, and we are very proud of it. The electrophysiology studies that we have carried out during this first year of the grant are very promising, since we have been able to observe how the mutation present in the PCCA patient significantly reduced the frequency of spontaneous activity, depressed cell excitability, presented differences in the potential action morphology due to alterations of the repolarization process, and presented delayed afterdepolarizations.

Our next step was to test the hypothesis that delayed afterdepolarizations are associated with an increased risk of

GRANT (CONT. PAGE 4)

arrhythmias and are due to severe alterations in cytosolic Ca2+ handling processes. For that aim, we first analyzed potential differences in the L-type Ca2+ current (ICaL) recorded in hiPSC-cardiomyocytes differentiated from the healthy donor and the PCCA patient. All these results demonstrate that the PCCA mutation significantly reduces de ICaL density suggesting that it severely affects in the Ca2+ handling process. We next wanted to determine putative effects of the PCCA mutation on the current generated by the Na-Ca exchanger (INCX), whose alterations have been associated with the appearance of delayed afterdepolarizations. These experiments will be completed in the next few months, allowing us to determine putative differences in the INCX due to the PCCA mutation. Additionally, to further investigate the mechanisms responsible for the alterations in the Ca2+ handling process, we will also analyze the late component of the sodium current (INaL), as it is extensively described the interplay between the magnitude of the INaL the intracellular Ca2+ concentrations and the delayed depolarizations.

Overall, the observed effects demonstrate that the presence of the PCCA mutation produces electrophysiological alterations that seem to be mediated by a mechanism that involves derangements in the Ca2+ handling process.

We think that our project is of significant clinical importance to understand the pathomechanisms underlying cardiac alterations in PA patients. Our results will provide the first experimental evidence of the altered ion currents in PA using a human cellular electrophysiological approach and would allow to get a mechanistic insight into the causes of the electrophysiological alterations observed in PA patients. The knowledge of the affected ion current and the underlying mechanism will open new avenues into the putative therapeutic interventions in these patients, with already existing tools (antiarrhythmic drugs) or with novel strategies in the context of personalized medicine.

We greatly appreciate the support and efforts made by PA families, PAF board members, volunteers, and donors to make this study possible. The funding we received has led to important advances in PA pathophysiology, and our aim is to continue this research in the near future.

REUBEN (CONT. PAGE 3)

Reuben, age 39

He is tube-fed all his nutrition, a mixture of Duocal, Anamix and Ensure and will snack on chips or Cheetos. He has taught me so much about patience and enjoying the little things in life and being satisfied with whatever comes our way.

Please feel free to contact me if you'd like to talk about our kiddos. I can be reached at Patt@CarolinaBehaviorandBeyond.com

We want to hear from you! Have a PA story to tell, event to promote or news? Spring newsletter submissions due by Feb. 15, 2024

Warriors Birthday Club

This year birthday cards will be made by students at Oak Lawn-Hometown Middle School and St. Linus for participating families. We are thankful they have volunteered to do it again this school year. Please sign up a patient or sibling for the Warriors Birthday Club at http://www.pafoundation.com/warriors-birthday-club/.
If you signed up last year, you will need to sign up again, so we have current information.

OFFICIAL
CHARITY PARTNER
tcs NEW YORK CITY MARATHON

NOVEMBER 05, 2023

TCS NEW YORK CITY MARATHON

Meet our 6 amazing 2023 NYC Marathon PA Runners. For more about their stories, check their fundraising pages.

John Moss, St Louis-Missouri, USA. "I am running for my twin boys Grant and Sebastian with Propionic Acidemia. My goals are to raise at least $3,000 for the PAF foundation so that PAF may continue to fund research for a cure, treatments and keep up the networking for families with so little information informed". https://fundraisers.hakuapp.com/john-moss

Sophia Kolodzinski, New York, USA. "If you know me, you know how excited I am to run my very first marathon!!! More importantly, I'm very honored to be running alongside a wonderful team of runners supporting an incredibly important cause, the Propionic Acidemia Foundation (PAF)! As someone who was born with a congenital condition, I am extremely connected to and so inspired by the strength and perseverance of the families and community affected by PA. I also feel so grateful and excited to be running alongside my close friends, Nina Korman and Emma Lewis, and with the PAF Marathon team!
https://fundraisers.hakuapp.com/fundraisers/e9345abc92a66cfebb67

Emma Lewis, New York, USA. "Hi everyone! My name is Emma Lewis and I am super excited to be running my first ever marathon with PAF. I grew up in the suburbs of NYC and currently reside in Manhattan. I've been running since high school and was motivated to run the marathon after cheering on fellow New Yorkers and friends in past NYC marathons. PAF's mission is inspiring, and I feel so grateful to now be running in the race as part of the PAF marathon team and to raise funds and awareness for PA".
https://fundraisers.hakuapp.com/emma-lewis

Lauren Piccolo-Ingram, New York, USA. "This year I am running the New York City Marathon to raise funds for the Propionic Acidemia Foundation (PAF) for my neighbors and friends Marisa Cotrina and Juan Carlos Lopez and their son Gabriel who suffers from propionic acidemia. Marisa has been a tireless fighter for PAF for years. I am joining the team she has organized to support her son and others who suffer from this rare metabolic disease." https://fundraisers.hakuapp.com/fundraisers/9d5baf45f2a609e41927

Nina Korman, New York, USA. "I'm so excited to be running the TCS NYC Marathon with PAF!! The strength and perseverance of the families and community affected by PA is so inspiring and I feel so lucky to be involved, spread awareness, and run for such an incredible organization. The countdown to November 5th has begun!"
https://fundraisers.hakuapp.com/fundraisers/64f0dca526b489d203fe

Mirella Siwik, Chicago-Illinois, USA. Mirella knows from own experience the hardship and the heartache that parents experience when trying to determine a diagnosis and treatment for their child. She also knows that Research is key. We are very happy to welcome Mirella to our PAF Runners team this year!
https://fundraisers.hakuapp.com/fundraisers/aacf5ad11592feac6134

PROPIONIC ACIDEMIA FOUNDATION

ANNUAL REPORT 2022-2023

By all definitions, the Propionic Acidemia Foundation has had a successful year for 2022-2023. PAF funded three research projects, held two conferences, board members attended two medical/dietetics conferences, created an *Emergency Handbook for Those with Propionic Acidemia/Methylmalonic Acidemia* and revised the patient Care Notebook.

We also had a more diverse fundraising effort this year. Eleven people ran in the for the New York City Marathon in November 2022 to raise money for PAF. Families and individuals held in-person and online fundraisers.

Impacts

- A new grant for $50,000, awarded to Pawel Swietach, University of Oxford, England for the project entitled "New paradigm in alleviating the cardiac consequences of propionic acidemia: diverting excess propionate towards the heart's beta-alanine store."

- A continuation grant for $50,000 awarded to Eva Richard, Centro de Biología Molecular “Severo Ochoa”, Madrid, Spain and Eva Delpón, Universidad Complutense de Madrid, Spain for the project entitled, "Elucidation of cardiac electrophysiology alterations in propionic acidemia: Towards the identification of targets for therapeutics."

- A continuation grant for $50,000 awarded to Guofang Zhang, PhD, Duke University “Propionyl-CoA and propionylcarnitine mediate cardiac complications in patients with propionic acidemia”

- Family conference held in March 2023 at Lurie Children's Hospital, Chicago, IL with 70 attendees.

- Family conference in July 2023 in Kidron, OH with 300 attendees. 85 EKGs, 78 ECHO 2D, 78 Strain Image, & 9 3D ECHO were given to participants for a study on PA and the heart.

- *My Care Notebook* & *Emergency Handbook for Those with Propionic Acidemia/Methylmalonic Acidemia* were displayed and distributed at PAF's conferences in Ohio and Illinois and is available to download..

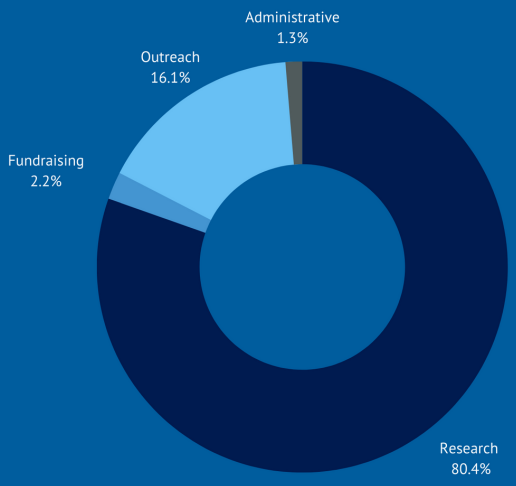
- Attended & exhibited at Society of Inherited Metabolic Disorders Annual Meeting in Utah, Abbott Metabolic Conference in Texas and Illinois PKU and Allied Disorders Annual Meeting in IL.

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.

FINANCIAL STATEMENT

Expenses



80.4% Research Grants & Registry	$152,752
16.1% Outreach Conferences, Educational Resources	$30,640
2.2% Fundraising	$4,117
1.3% Administrative	$2,496
Total Expenses	**$190,005**

Income

Cash assets 8/1/2022	$455,691
Cash assets 7/31/2023	$438,080

Donations	$165,533
Interest Income	$6,861
Total Income	**$172,394**

Board of Directors

Jill Chertow, President
Brittany Smith, Treasurer
Angela Waits, Secretary
Maria L. Cotrina
Veronica Lopez

Propionic Acidemia Foundation

PO Box 151
Deerfield, IL 60015-4421
1-877-720-2192
www.pafoundation.com

PAF ACTIVITIES AND FUNDRAISING SPOTLIGHT

UPCOMING EVENTS

- 10/14 Gwen & Allison for a Cure Fall Festival - OH
- 10/21 Exhibiting at PKU & Allied Disorders Meeting - IL
- 11/5 NYC Marathon - Support the PARunners!
- 11/17-11/18 Kish Valley Meeting - Belleville, PA (more info to follow - Day 1 - Propionic Acidemia)
- 11/28 Giving Tuesday

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: Harper Anne Asher, Gabriel Lopez, Sebastian and Grant Moss, Trent McKinley,
- In Memory Of: Harvey Crouch, Jordan Franks, Tallina Smith, Fred Uhlmann

FACEBOOK: Thank you to all of our Facebook Fundraisers and people that donated to their fundraising pages for birthdays, #GivingTuesday or just because: Linda Chell, Amanda Jackson, Sonia Phillips, Iris (Jack Trimmell, Jr.), Lisa Shutts-Mash, Elfie Sirett, Danny Denice Harris, Lisha Li

STOCK DONATIONS: PAF accepts stock donations. Please email paf@pafoundation.com with any questions.

MATCHING DONATIONS AND 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; selling on Ebay and setting up Facebook Fundraising Pages and designating Propionic Acidemia Foundation as your charity. Every dollar helps.

Giving Tuesday - November 28, 2023

Every act of generosity counts!

GivingTuesday was created in 2012 as a simple idea:
a day that encourages people to do good.

Ways to Participate

1. Donate to PAF (every dollar counts).
2. Set up a fundraiser (Facebook fundraiser, restaurant night, or home party).
3. Volunteer virtually or in person.
4. Show gratitude to your medical team, school team or other important people in your life (thank you cards, sweet treats, etc.)
5. Spread awareness by sharing your story or telling people about propionic acidemia.

CLINICAL TRIALS

HERO Clinical Trial Recruiting Participants

The **HERO** (**H**elp **R**educe **O**rganic **A**cids) clinical study, sponsored by HemoShear Therapeutics, is recruiting participants at 13 leading research hospitals across the United States.

"We are grateful to the patients and families who are participating in our clinical trial, and happy to share that there are still opportunities to join the study," says Pat Horn, MD, PhD, Chief Medical Officer at HemoShear. "We have many locations across the country, and transportation can be provided to make the study more convenient for families to join."

HERO is actively recruiting children and adults with MMA (mutase deficient) and PA aged 2 and older who meet the study criteria. While in the study, participants can continue to take their medications, including carnitine. More information about the HERO study is provided at MMA-PAHero.com or clinicaltrials.gov.

Study Assessing Potential New Treatment

HemoShear is developing the investigational drug HST5040 as a potential treatment for MMA and PA. HST5040 is a liquid that is taken at home twice daily by mouth or through a gastric or nasogastric feeding tube. The HERO study is designed to assess how HST5040 acts in the body, if it causes side effects and whether it works to reduce harmful toxins in the body and help people with MMA or PA feel better.

Find a Site Near You

HERO is being conducted at many children's hospitals across the United States, as well as Australia and Saudi Arabia. Ask your doctor about whether you could be a candidate to participate.

Transportation can be provided to travel to the study sites and stipends are available to cover meals and other study-related expenses. Some visits can also be performed in your home. All study drugs, study visits and assessments will be provided at no cost.

Learn more at MMA-PAHero.com

The safety and effectiveness of HST5040 for the treatment of MMA or PA have not been established.

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

SEARCHING FOR A CURE
HOPE FOR OUR CHILDREN

PROPIONIC ACIDEMIA FOUNDATION

Sponsor Partners

moderna

HemoShear Therapeutics
Recordati Rare Diseases
Nutricia

Newsletter Committee

Julie Howard
Jennifer Mouat

Birthday Club Volunteers

Heather McCarthy
Oak Lawn Hometown Middle School

Connect with us:

pafoundation.com
paf@pafoundation.com
Patient Registry: paregistry.org
877-720-2192
/propionicacidemia
/company/propionic-acidemia-foundation
/propionic_acidemia/

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, P.O. Box 151, Deerfield, IL 60015-4421. If you would like to be removed from our mailing list or receive the newsletter via email, please contact us.