

# ANNUAL REPORT 2020-2021



**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.

## 2020-2021 Financials

### Revenue:

Contributions	\$139,492
Interest	\$650
<b>Total Income</b>	<b>\$140,142</b>

### Expenses:

Program services	\$174,565
Management & General	\$1,978
Fundraising	\$221
<b>Total Expenses</b>	<b>\$176,764</b>
Cash Assets 8/01/2020	\$468,376
Cash Assets 7/31/2021	\$431,754

## Board of Directors

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

### Letter from the Board of Directors

We are happy to share with you our annual report.

This past year has been one like no other; having to handle the day-to-day life with Propionic Acidemia can be very difficult, but add in a pandemic and it has stretched families and their providers.

As an organization, we have had to make adjustments. PAF granted research grant extensions to our grant recipients as labs were closed. We had to postpone an in-person conference we were planning, but were able to hold a virtual conference in October 2020 with international participation.

We hosted virtual events, including dance parties and art classes over the past year. We also hosted a PA chat for families to share their experiences. Collaborative meetings with other rare disease groups over Zoom provided information on low protein diets, advocacy, and clinical trials. We thank you for your support.

**Board Disclosure:** Donations made by board members totaled \$600.

# PROGRAM ACCOMPLISHMENTS

## Grants Awarded

- Awarded \$49,953 grant to Pawel Swietach, DPHIL, at the University of Oxford, England as a new grant for the project entitled: “Aberrant protein propionylation and distinct histone marks in propionic acidemia: new disease mechanisms and risk factors for cardiac disease.”
- Awarded \$50,000 grant to Sander Houten, Ph.D. & Robert J DeVita, PhD, at the Icahn School of Medicine at Mount Sinai, New York, USA as a new grant for the project entitled: “Substrate reduction as a novel therapeutic strategy for propionic acidemia.”
- Awarded \$1,890 grant to Community Health Clinic in Topeka, Indiana, USA to perform genetic testing for c.1606A>G in the Old Order Amish (OOA) community.
- Awarded \$51,500 grant to Guofang Zhang, PhD, Duke University, Durham, North Carolina, USA as a continuation grant for the project entitled: “Propionyl-CoA and propionylcarnitine mediate cardiac complications in patients with propionic acidemia”

## Support Activities

- In October 2020, PAF held a virtual “Family Day” with speakers from the National Institutes of Health, Bethesda, Maryland, USA and Boston Children’s Hospital, Boston, Massachusetts, USA. Video recordings of two of the speakers are available for families to view on-line.
- Distributed fall and spring newsletters to affected families, clinicians, and donors
- Coordinated the Propionic Acidemia Foundation’s Warrior Birthday Club in which students and staff at Oak Lawn Hometown Middle School and St. Linus School, Illinois, USA make birthday cards for those affected by PA, their siblings, and children.
- Hosted “Fun times with Shayna”, virtual dance parties and art classes over Zoom.

## Registry

114 Participants in the PA International Patient Registry. The Registry is an IRB-approved research project and the data collected will help characterize the condition of people living with PA.

## Publications on PAF funded grants

Alonso-Barroso E, Perez B, Desviat LR, Richard, E. (2021) Cardiomyocytes Derived from Induced Pluripotent Stem Cells as a Disease Model for Propionic Acidemia. *International Journal of Molecular Science*. 22:1161. doi:10.3390/ijms22031161

Fulgencio-Covián A, Álvarez, M, Pepers, BA, López-Márquez A, Ugarte, M, Pérez B, van Roon-Mom WMC, Desviat, LR, Richard, E. (2020) Generation of a gene-corrected human isogenic line (UAMi006-A) from propionic acidemia patient iPSC with an homozygous mutation in the PCCB gene using CRISPR/Cas9 technology. *Stem Cell Research*. 49:102055. doi:10.1016/j.scr.2020.102055

Park KC, Krywawych S, Richards E, Desviat, LR & Swietach. (2020) Cardiac Complication of Propionic and Other Inherited Organic Acidemias. *Frontiers in Cardiovascular Medicine*. 7:617451. doi:10.3389/fcvm.2020.617451

Saleemani H, Egri C, Horvath G, Stockler-Ipsiroglu S, Elango, R. (2021) Dietary management and growth outcomes in children with propionic acidemia: A natural history study. *JIMD Reports*. doi:10.1002/jmd2.12234

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