

Propionic Acidemia Foundation

VOLUME 1, ISSUE 23

FALL 2017

Warrior Wisdom Conference

On June 8th, 9th, and 10th the Propionic Acidemia Foundation held it's Warrior Wisdom Conference in Deerfield, Illinois. Attendees included fifteen



families affected by PA; physicians, researchers, and other providers; representatives of medical foods companies; and local support organizations. Families from as far as California, Texas, and New York made their way to Deerfield to be in attendance.

On the evening of the 8th, participants gathered for a reception with Chicago-style snacks including popcorn and root beer. The 9th and the morning of the 10th was filled with presentations. Dr. Barbara Burton gave an overview of PA. There were three presentations by researchers at NIH, Dr. Charles Venditti, Dr. Iriini Manoli, and Dr. Oleg Shchelochkov on the progress of the natural history study of PA, different dietary management of PA, and research on MMA and how that translates to information on PA. On behalf of the collaborative effort of the Genetic Metabolic Dietitians International (GMDI) and the Southeast Regional Genetics Network (SERN), Keiko Ueda and Amie Thompson gave updates on their Nutrition Management Guidelines for PA and asked for input on developing their FAQs (see page 2). Dr. Jan Kraus gave an update on his research with enzyme replacement. Dr. Nicholas Ah Mew relayed information on the N-Carbamylglutamate trial for those with PA. Dr. Gerard Berry discussed liver transplantation. Dr. Hilary Vernon provided information on her research on cellular pathways in PA. Dr. Holmes Morton was not able to attend; however, he was able to give his presentation remotely on the (continued page 5, bottom)

PA Registry

Help move research forward for propionic acidemia. Participate in the Propionic Acidemia International Registry. As of October 1st, there are 56 participants. For more information on joining the registry, or to update your information, go to www.paregistry.org.

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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.

ABBOTT NUTRITION CONFERENCE

16TH Abbott Nutrition Metabolic Conference Advances in Management of Inherited Metabolic Disorders

March 23-25, 2017

By Marisa Cotrina, PAF board member

Summary

Abbott Nutrition invited PAF to attend the annual nutrition metabolic conference this past March in the Isle of Palms, South Carolina.

Two talks were of particular interest for propionic acidemia (PA) patients. Dietitian Elaina Jurecki presented the new evidence-based nutrition guidelines that were just finalized for the PA community; an initiative that started several years ago in collaboration with some of the best dietitians in the country and that is finally panning out in these series of guidelines that, hopefully, will make management of PA more uniform and effective in the USA.

For those dietitians new to PA, Dr. Loren Pena presented the metabolic bases for understanding and treating PA. I was very grateful to see how Dr. Pena kindly advocated for the PA community and highlighted the role PAF and the patient organizations play in supporting new research initiatives to advance PA therapies.

The topic of liver transplantation and how effective it may be in reversing and/or preventing metabolic damage was discussed in the context of MSUD with dietitian Melanie Reeves.

It was clear from this talk the urgency to publish more data on liver transplantation from all the transplant units in the country so that patients and clinicians can make a more educated decision about the risks and benefits of this approach for the long-term management of metabolic disorders.

Lastly, Dr. Mark Korson moderated an exercise with team ChoP and team Oregon to discuss two crucial topics in our community: 1) can metabolic disorders be managed without medical formulas?, and 2) when is it cost effective and ethically appropriate to include metabolic disorders in NBS? Excellent presentations from both teams and fantastic discussion.

Dietitians are a critical line of defense in the chronic management of PA and we need to listen and work with them as much as with our medical doctors. Overall, a great learning opportunity for PAF and our community. (For a complete report, go to <http://www.pafoundation.com/paf-attends-16th-abbott-nutrition-metabolic-conference/>)



NEXT STEPS FOR THE PA NUTRITION GUIDELINES

Elaina Jurecki MS, RD and Keiko Ueda, MPH, RD, PROP Workgroup Chairs

The Propionic Acidemia (PROP) workgroup has successfully completed the development of the Nutrition Management Guideline for PROP, a collaborative effort between Genetic Metabolic Dietitians International (GMDI) and the Southeast Regional Genetics Network (SERN)*. Any recommendations, such as these, are not effective unless there are tools to help implement them. The next step is to develop a tool kit to help support this endeavor. The PROP tool kit will provide practical examples for metabolic dietitians and other clinicians to apply the Nutrition Guidelines in caring for their patients living with propionic acidemia. We aim for the PROP toolkit to be available by Spring 2018 on the Nutrition Guidelines webpage via: southeastgenetics.org or gmdi.org. We also plan for the PROP Nutrition Guideline manuscript to be submitted for publication in a peer-reviewed medical journal by the end of this year.

In February 2017, the Nutrition Guidelines Core group and PROP Workgroup Chairs had a meeting with patient advocates representing PAF and the Organic Acidemia Association (OAA). It was determined that a Summary Sheet with an accompanying Frequently Asked Question (FAQ) sheet would be helpful for patients, families and caregivers living with PROP. The Summary Sheet is a one-page list of things to consider and discuss with your metabolic team. This summary is not meant to provide medical advice. Some items addressed in the summary include when to contact your metabolic clinic, certain medications to consider, suggestions for sick day instructions, and what laboratory and physical assessments should be followed. The Frequently Asked Questions (FAQ) sheet includes questions addressing such things as: Nutrient Intake (i.e. How much protein can I handle?) and Supplements (i.e. Do I need isoleucine or valine supplements?). Other topics are Blood Monitoring, Illness, Liver Transplant and Pregnancy. Both the Summary and FAQ... (continued on page 3)

RESEARCH GRANT UPDATES

PAF Awards \$43,645 Research Grant

Hilary Vernon, MD, PhD, Johns Hopkins University
“Targeting Serine and Thiol Metabolism in Propionic Acidemia”

In the research project, entitled **“Targeting Serine and Thiol Metabolism in Propionic Acidemia”**, we will define the role of novel pathways of cellular dysfunction in Propionic Acidemia. It has been known for several decades that the genetic cause of Propionic Acidemia is an abnormality in one of the genes encoding for propionyl-CoA carboxylase, leading to accumulation of propionic acid and its metabolites. However there is still a limited understanding of how this causes cellular dysfunction.

In our preliminary work, we defined novel differences in the way that cells affected with Propionic Acidemia produce defenses against oxidative stress, and in the way that these cells utilize serine, a central amino acid for cellular growth. In this current research project, we will use a new cell model of propionyl-CoA carboxylase deficiency developed in our laboratory to fully define these pathway differences, and how they alter the response to stress. We hope that in defining these new pathways of cellular dysfunction in Propionic Acidemia, we will offer new areas for potential therapeutic intervention and therapeutic monitoring. We are excited to begin this research, and are grateful to the Propionic Acidemia Foundation for funding this research.

DR. HILARY VERNON
 ASSISTANT PROFESSOR OF GENETIC MEDICINE
 MCKUSICK-NATHANS INSTITUTE OF GENETIC MEDICINE
 JOHNS HOPKINS UNIVERSITY

PAF Awards \$50,000 Research Grant

Nicola Brunetti-Pierri, MD, Fondazione Telethon, Italy
“Novel Therapies for Propionic Acidemia”

In June 2017, PAF awarded a \$50,000 grant. Propionic acidemia (PA) is an inborn error of metabolism caused by deficiency of propionyl-CoA carboxylase (PCC). There is no cure for PA and available therapies are often unsatisfactory. Elective liver transplantation is being increasingly used to reduce metabolic crises and temper some of the most severe symptoms of the disease. However, liver transplantation is invasive, has high morbidity, and requires long-term immunosuppression. Therefore, more effective and safer therapies for PA are highly needed. We generated a PA model in the medaka fish (*Oryzias latipes*) that recapitulate the pathological and biochemical abnormalities observed in human patients, and has reduced locomotor activity and survival. As disease model, the fish has several advantages including easy handling, low costs, large number of progeny per generation, a relatively short generation time, and a small and well-draft genome. We propose to perform a high-throughput drug screening in this animal model using the reliable and clinically relevant endpoint of locomotor activity as first-tier read-out. Positive hits will be confirmed for their effect on biochemical abnormalities and survival.



NEXT STEPS (CONTINUED FROM PAGE 2)

Sheets are resources that you can bring to clinic appointments to help identify topics to discuss about your/your child's care. They can also be helpful in informing family and friends about PROP.

In June 2017 at the PAF Warrior Wisdom Conference in Deerfield, IL; the initial drafts of the PROP Summary and FAQ sheets were presented by Keiko Ueda and Amie Thompson, RD, LD, PROP Workgroup member to parents, adults living with PROP, PROP medical experts and researchers. We received valuable feedback from participants that has been shared with our Nutrition Guidelines Core group to help update and improve the Summary and FAQ Sheets. We hope to finalize these tools and have them available on the PROP Nutritional Guidelines website in the near future. We greatly

appreciated the opportunity to attend the 2017 PAF conference and network with families, PROP dietitians, medical experts and researchers. We gained valuable insights from parents and patients living with PROP from their questions and challenges raised by implementation of the PROP Nutrition Guidelines.

Consumer input is very helpful as we continue to develop tools to optimize nutritional management for individuals living with propionic acidemia (PROP/PA). If you would like to review and provide feedback as well as ask questions about the PROP Nutritional Guidelines, please visit the PROP Nutritional Guidelines webpage and fill out the feedback and comments form. Thank you!

*GMDI/SERN Nutrition Guideline Development is a HRSA Supported Partnership — Grant #UH7MC3077

HEATHER'S CONFERENCE EXPERIENCE

What I Learned From Attending a Propionic Acidemia Conference

By Heather McCarthy

I'm sitting in a conference room listening to researchers, doctors, and dietitians challenge each other. They're questioning each other's studies, pressing to gain more information, debating on best practices. This would normally sound like a nightmare, but I'm in heaven.

As a mom to two children with propionic acidemia, it's music to my ears to sit in a room full of specialists wanting to find the best treatment, solutions, and care for my children and our rare community.

I had the privilege of attending the Propionic Acidemia Foundation's Warrior Wisdom Conference. When you're part of a rare community, you don't always have the opportunity to meet likeminded people. Today I sit with 15 different affected families from 17 states and Canada. The room is full of doctors, dietitians, nurses, nurse practitioners, researchers, school support, and genetic counselors from 10 leading institutions.

From this conference, I learned that we may be small, but we are mighty. It's understandable that researchers want to find a cure for the most common diseases. If you cure something that affects many, your cure has a greater impact. So to find so many people listening, challenging, and debating, I could not be happier. Research shows only one in 100,000 people in the U.S. are affected by propionic acidemia. While that puts us in a rare category, it also helps us to become even mightier. Every time we can get doctors and researchers to communicate, connect, and debate with each other, we become stronger.



Every time we can bring families together to talk to each other, share stories, and help each other, we become mightier.

Every time we can challenge nutritional guidelines, therapies, and funding, we become more powerful.

From this conference, I learned that we are important. Moms and dads, siblings, and other family members in the rare community always find each other important. This conference showed we are important to the researchers. We are important to the doctors. We are important to the nutritionists, to the genetic counselors, the nurses, to the undiagnosed, to the future medical field, and to the children who may be born with propionic acidemia.

Finally, I learned there is hope. I learned that together we can expand newborn screening processes, work to get coverage for medical formulas and foods, and begin drug trials and new studies. We can expand patient registries and open additional grants for new studies and research projects.

If you belong to a rare community, you are not alone. You can be rare and mighty at the same time. You are unique and you are important. Have hope!

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. To find out more, visit <http://www.pafoundation.com/>.

If you would like to read more articles by Heather McCarthy, visit themighty.com.

RACHEL'S STORY



My name is Rachel. I am 40 years old and have propionic acidemia. I live with my mom and dad. Three days of the week I work at a work center for the disabled and on 2 days I go to work at the Painted Turtle. I love both my jobs but the Painted Turtle is my favorite. I get to paint and do other artwork that is sold to the public.

I love to read and go on my computer. I attend events at my local Special Recreation Program. We go bowling, have dances and attend plays.

My favorite vacations are at Disney World and the Disney Cruises. Disney is very good about helping me with my special diet.

THANK YOU FROM NIH CAREGIVER STUDY

The Caregiving Study team at the National Institutes of Health would like to extend a big thank you to all the families that have participated in the study thus far! We were excited to attend the PA conference in June, giving us the unique opportunity to meet with families, and collaborate with researchers and healthcare professionals all dedicated to supporting families affected by PA. In addition, we also asked families attending the PAF conference to fill out a short survey, in which we received valuable information regarding how far families have to travel to receive metabolic specialist care, their experiences in emergency room departments, and following prescribed medical diets.

The Caregiving Study is a project focused on learning to better understand the experience of parents and caregivers who care for children affected by inborn errors of metabolism, including PA. Parents and caregivers who share their experiences help us understand how the social environment influences how family members cope, which will ultimately help us learn how we can better support families facing the challenges associated with metabolic disorders.

As of August 2017, we have had over 90 families participate. Our participants include parents, other relatives, and caregivers of children with inborn errors of metabolism or undiagnosed diseases. These families come from all over the world.

Our families have given us insight into the complex nature of the difficulties that caregivers and parents encounter, including struggling to find dependable respite care, the daunting nature of dealing with insurance companies, and the overall financial burden of medical costs. When asked about coping, many have shared that caregiving has caused a positive shift in their perspective, such as helping them and their family members become more empathetic toward others with special needs. Our study team looks forward to hearing more perspectives about caregiving experiences as we continue to enroll families in all parts of the study. If you have any questions about our study, please contact the study team:

Kate Marcum
Clinical Research Coordinator
National Institutes of Health
301-219-3394
Kathleen.marcum@nih.gov



WARRIOR WISDOM CONFERENCE (CONTINUED FROM COVER)

Amish/Mennonite variant and outcomes with a new treatment protocol. Families also had the opportunity to learn more about current studies and meet with representatives from the Caregiving Study team from NIH (see page 5). The presentations shared new information and sparked many important discussions on propionic acidemia.

This event would not have been possible without the fundraising efforts of Deerfield High School and sponsorship from Nutricia North America, Recordati Rare Diseases Group, Vitaflo, Cambrooke Therapeutics, PKU Organization of Illinois, and Mevalia.

Student volunteers from Deerfield High School as well as the Chertow, Rosenson, and Schneeweis families provided activities and childcare for children affected by PA and their siblings. Thank you so much!!



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.

PAF EVENT & FUNDRAISING SPOTLIGHT

PAST EVENTS & CAMPAIGNS:

- **Matz Family Garage Sale** - \$1125
- **12th Annual Tailgate Party & Corn Hole Tourney** - \$10,000+

UPCOMING/ONGOING EVENTS

- **Fundraiser Crop, October 21, Galena, OH**
For info: im2alesmom@yahoo.com

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.
- **Internet:** Thank you for using Igive, Goodsearch and AmazonSmile and designating Propionic Acidemia Foundation as your charity. Every dollar counts.

MAKE YOUR ONLINE SHOPPING COUNT WITH:

- Igive
- Goodsearch
- AmazonSmile

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: Mr. and Mrs. Gene Koehl's 50th Anniversary, Kate Lowry, Gabriel Lopez, Talli Smith

In Memory Of: Vincent Franze, Jordan Franks

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.

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

WARRIORS BIRTHDAY CLUB

For the 2017-2018 school year, PAF is partnering with Oak Lawn Hometown Middle School. The students will be making cards for individuals with propionic acidemia and their siblings. If you would like to participate, please fill out the online form at the link below by November 15, 2017.



If you are located outside of the United States, in lieu of cards, we will be sending emails. You can join at <http://www.pafoundation.com/warriors-birthday-club/>

SIGN UP BY NOVEMBER 15!!

PAF Puzzle



Cut out the puzzle pieces on the white lines and reassemble.

ANNUAL REPORT FY 2016-2017

FINANCIAL REPORT

Revenue:

Contributions:	\$61,242
Fundraisers:	\$178,368
Royalty Income:	\$6,400
Program Service:	\$2,313
Interest Income:	\$1,671
Total Revenue:	\$250,318

Expenses:

Programs:	\$92,951
Fundraising:	\$449
Management & General Expenses:	\$3,447
Total Expenses:	\$96,846

Cash Assets 8/1/2016: \$226,749

Cash Assets 7/31/2017: \$380,221

Board Disclosure: Donations from Board Members totaled \$1,730

PROGRAM ACCOMPLISHMENTS

- Hosted PAF Warrior Wisdom Conference, June 8-10th
- Awarded two research grants
- Distributed fall and spring newsletters to affected families, clinicians, and donors
- Participated in Management Guidelines Consumer Toolkit Meeting
- Attended and exhibited at 16th Abbott Nutrition Metabolic Conference

BOARD OF DIRECTORS/OFFICERS

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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? Spring newsletter submissions due by March 1, 2018!

Help Us Find the Cure!

Name _____

Please send an acknowledgement to:

Address _____

Name _____

City, State, Zip _____

Address _____

Phone _____

City, State, Zip _____

E-mail _____

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

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needed!**

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