



National  
**PKU**  
Alliance

research | education | support | advocacy

## 2016 Annual Report



### A Message from the Executive Director

As I reflect on the last eight years since the founding of the National PKU Alliance, I am continually amazed and humbled at how much we have been able to accomplish together as a community.

We all want adults and children living with PKU to have better lives. With your help, we have invested more than \$2 million in research seeking better treatments, better understanding, and paths to a cure. These investments have led to new knowledge discovery of how PKU affects the white and gray matter structures in the brain, helped to create a new animal model for PKU to better understand the course of the disorder and potentially speed up the drug development process, and led to new careers for bright and talented young scientists to pursue new therapeutic approaches to treat PKU.

Our conferences have brought together close to 2,000 families and researchers to form lasting friendships, support systems and key learnings in how to best manage PKU. Our maternal PKU mentoring program provides critical support to adult women who are pregnant to give birth to healthy

babies. Our military families now have the medical foods coverage they need to treat PKU.

However, we need to do more. The challenges before us continue to be great as 61.5% of adults and 25.5% of children are not currently within the recommended Phe level range and 91% say the development of new treatments is important to them. Many still cannot get the medical coverage they need to treat PKU even though all states are required to screen for the disorder. We live in an exciting and innovative time – there are now more treatments in development for PKU than in any other time in history. Together, we can and will continue to make significant gains for our loved ones with PKU. Together, we will accelerate new treatments and find a cure.

*Christine S. Brown*

Christine S. Brown, MS  
Executive Director

#### Save The Date! Lifting the Limits for PKU

April 29, 2017  
Buffalo, New York

#### PKU Advocacy on the Hill Days

May 22-23, 2017  
Washington, D.C.

#### Creighton Farms Invitational

June 24-26, 2017  
Middleburg, Virginia

#### Lifting the Limits for PKU

November 4, 2017  
Princeton, New Jersey

#### 2018 NPKUA Conference

July 5-8, 2018  
Atlanta, Georgia

Find more information  
on these events at [npkua.org](http://npkua.org)

## 2016 NPKUA Research Awards

**Dr. Juan Cabrera-Luque** at George Washington University is looking at ways to establish a novel in vitro model to study the effects that high blood phenylalanine concentrations have at the human blood brain barrier level.

**Dr. Katherine Durrer-Deming** at the University of North Texas is researching the ability of a genetically engineered probiotic to lower blood Phe levels in PKU mice that could result in a new treatment for PKU.

**Dr. Cary O. Harding** at the Oregon Health & Science University is researching the use of gene therapy using CRISPR-Cas9 reagents to directly correct the PAH gene in PKU mice, resulting in a possible cure for PKU.

**Dr. Roberto Gramignoli** at the Karolinska Institutet in Stockholm, Sweden has described the importance and peculiar characteristics of amnion epithelial cells (AEC) in the human placenta to explore cellular therapy in PKU.

**Dr. Robert Nicholls** at Children's Hospital of Pittsburgh of UPMC developed reagents for genome editing of PAH in pig embryos to produce a miniature pig model of PKU.

**Dr. Dong Yizhou** at Ohio State University is researching CRISPR-Cas9 technology to correct the PAH gene to produce a functional PAH protein and recover the metabolic process, resulting in a possible cure for PKU.

**Dr. Paulo Roque Lino** at iMed.Ulisboa (Research Institute for Medicines, Faculty of Pharmacy, University of Lisbon, Portugal) was awarded a fellowship for the second year to focus on the development of an enzyme reposition therapy approach to PKU.



The NPKUA held another outstanding PKU conference in Indianapolis, Indiana, July 28-31. The largest NPKUA conference to date, over 600 attendees including families, researchers and industry professionals, networked, shared and learned updates on the latest NPKUA funded research projects, as well as new studies and information on other research. An update on the Home Phe Monitor was also provided.

Through various educational offerings, adults and teens came away with new information on ground breaking research, everyday tips to manage PKU better, important insurance information, fundraising and awareness activities, and much more. Children with and without PKU enjoyed time with friends in the Kids Zone, took a field trip to the Children's Museum and were involved in age appropriate educational sessions with dietitians.



**Thank you to our sponsors for supporting another important and largest PKU conference in the world! Thank you to the volunteers that helped make it a success!**

## Taking the Lead for PKU in Indianapolis



The NPKUA had the privilege of honoring a remarkable leader in the PKU community. Dr. Rani Singh received the PKU Hero Award for her work as an established leader and a trusted dietitian to PKU patients for more than 25 years and a national PKU champion in public policy and maternal PKU.

## Asbjørn Følling Society (AFS) Launched

At our conference, the NPKUA introduced the Asbjørn Følling Society to recognize the most generous annual donors to the NPKUA who make a gift of \$1,000 or more during a calendar year. The society is named in honor of Dr. Asbjørn Følling, the physician who not only discovered PKU in two Norwegian children, but then went on to examine other children living in mental institutions that led him to further characterize PKU. Qualifying founding members in 2016 donated more than \$500,000 through annual giving and Lifting the Limits for PKU events to benefit the NPKUA's work in research, education, and advocacy.

# Lifting the Limits for PKU Across the Nation



We had a very successful 2016 hosting Lifting the Limits events across the nation, starting on the east coast in Boston, MA, to Middleburg, VA, then journeyed over to the pacific coast in Seattle, WA. To start the year's events off, Boston hosted a very successful gala in May, bringing awareness to a new level and raising an outstanding \$400,000. Next, a first-ever golf tournament was held in Middleburg, Driving for a Cure. The Pro-Am Golf tournament raised over \$200,000 and opened the door to new opportunities for PKU fundraising. The final event of 2016 was a gala held in October at the Museum of Flight in Seattle, and soared to new heights raising funds exceeding \$550,000. These important events have significantly built the NPKUA Fund for PKU research and is helping to achieve our goals.

*Thank you to our co-chairs and their families for dedicating their time and commitment to help us Lift the Limits for PKU in your cities!*

*Thank you to volunteers, sponsors and donors for investing in the next generation of therapies and a cure!*

## Advocating for Research at the National Level

In the last year, a delegation from the NPKUA met with our federal stakeholders at both the National Institute of Health and the FDA to discuss the importance of developing new treatments for PKU, investing in PKU research and why the current treatment options available are not good enough to meet the needs of our community. In addition, the delegation brought the

power of the patient voice to these meetings by sharing the challenges of treatment adherence, the reality of maternal PKU syndrome and what symptoms of PKU are the most important to address in the development of new innovative and optimal therapies for PKU. These meetings are the first step in developing closer relationships with our partners at the federal level.

## Growing the Maternal PKU Program

In 2016, the NPKUA recruited six additional mentors to the Maternal PKU Mentor Program and hosted a two day training session following the conference in Indianapolis. The program supports women with PKU who are pregnant or considering becoming pregnant and establishes a mentor-mentee relationship between two women with PKU; one being pregnant or planning to become pregnant and the other having already experienced a successful pregnancy. The program currently has 10 mentors and 13 mentees. To date, the Maternal PKU Mentor Program has had 21 successful pregnancies. More information can be found at [www.adultswithpku.org](http://www.adultswithpku.org).

*"NPKUA empowers women to take control of their diet in order to have healthy pregnancies from the pre-conception diet, to the pregnancy itself and into the post-partum phase. Being able to teach women to advocate for themselves through sharing my experiences, passing on information so that they become more knowledgeable—and no matter what stage they are in—showing them the importance of diet for a healthy mom and healthy baby is one of the best gifts a woman with PKU can receive."*

*~Margaret, Mentor*



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After more than two years of research and planning the NPKUA and National Organization for Rare Disorders (NORD) launched the PKU Patient Registry globally in early 2017. This patient centered registry, open to individuals with PKU or a parent or guardian, provides the opportunity to collect information that can be used by clinicians and researchers to improve PKU treatment and accelerate research. The registry will provide an enriched understanding of the experience of living with PKU every day. Visit the registry at <http://pku.iamrare.org> to learn more and to enroll.

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\*not serving in 2017

NPKUA's audited financials are available by request by contacting Christine Brown at: [christine.brown@npkua.org](mailto:christine.brown@npkua.org) or 715-437-0477.