SAVE THE DATE!

2018 NPKUA CONFERENCE
Atlanta, GA - July 5-8, 2018

LIFTING THE LIMITS FOR PKU EVENTS
DENVER, CO
May 5, 2018

CREIGHTON FARMS INVITATIONAL | MIDDLEBURG, VA
June 23-25, 2018

CLEVELAND, OH
October 6, 2018

NEW YORK, NY
October 25, 2018

A MESSAGE FROM THE NPKUA PRESIDENT

As the mother of two children with PKU, I dream of a future where there is a cure. I know that we are making a difference in the lives of those with PKU while we all work towards achieving that dream. Ten years ago, a national patient organization that could represent the voice of PKU and meaningfully impact the future for PKU was still a dream. Then a dedicated group of families, friends, neighbors, colleagues, organizations, and others came together to make that dream come true. As we celebrate the tenth anniversary of the foundation of the National PKU Alliance, it's amazing to see how far we've come, and the progress we have made in reaching that ultimate dream of a cure. There are new treatments in the pipeline and the NPKUA continues to fund promising research that may someday result in a cure. We have a national patient voice and have become the preeminent representative of the PKU community. As we hold our “Advancing the Dream” conference in Atlanta this summer, the future for PKU is bright. With your continued support, we can continue to advance the dream of a cure for PKU.

Amy Oliver, JD
NPKUA President

Find more information on these events at www.npkua.org
Dr. Katherine Durrer, University of North Texas, through a fellowship, 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. Roberto Gramignoli, Karolinska Institutet in Stockholm, Sweden, is focused on cell-based therapies to correct metabolic defects and is progressing in research with cell transplantation, resulting in a possible cure.

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

Dr. Eileen K. Jaffe, Fox Chase Cancer Center, is studying how structure changes in PAH ensure the control of Phe concentration, which could result in a new treatment for PKU.

Dr. Robert Nicholls, 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 and is establishing offspring to better understand biomedical basis and allow for testing of new therapies for PKU.

Dr. Francjan van Spronsen, University of Groningen, Netherlands, is studying why high phenylalanine levels affect each person differently.

Dr. Susan Waisbren, Boston Children’s Hospital, is using new techniques to define and close the gap on how PKU affects the brain to better evaluate new treatments for PKU.

Dr. Dong Yizhou, Ohio State University, is also 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.

NPKUA FELLOWSHIP PROGRAM SUCCESS

Dr. Kristen Skvorak was one of the first NPKUA Fellows. The NPKUA Fellowship Program encourages bright post-doctoral researchers to pursue careers in metabolic diseases. Dr. Skvorak received funding from the NPKUA beginning in 2012. Five years later she is now employed with Codexis, which is developing an orally delivered enzyme as a possible new treatment for PKU.

NPKUA BRINGS PATIENTS TO THE FDA

The NPKUA met with FDA officials in October as part of the review process for a potential new treatment for PKU adults that is currently under review. Six adult patients participating in the clinical trials shared their personal stories with the FDA, while NPKUA Executive Director Christine Brown discussed the challenges that many adults face in keeping their Phe levels within the recommended range. 91% of adults and families have reported that the development of new treatments for PKU is important. Brown also shared with FDA officials the symptoms of PKU that were the most important to patients in the development of new therapies: drop in blood Phe levels; improvement in executive function skills, such as the ability to plan, organize and prioritize; and improvement in depression, anxiety and overall mood. Lastly, Dr. Cary Harding and Dr. Barbara Burton discussed with FDA reviewers their experience as principal investigators participating in the trial. The NPKUA will continue to engage and grow our partnership with the FDA to ensure that the patient experience remains the focal point of future clinical trials.

PKU PATIENT REGISTRY LAUNCHED

A big THANK YOU to every PKU patient, parent or caregiver who made the first year of the PKU Patient Registry so successful! Since its launch in January 2017, over 550 have enrolled in the PKU Registry and more than 60% have entered information into their surveys. Registry participants represent PKU across the globe, residing in 5 different countries and 41 states in the U.S. Our commitment to collecting the best data possible to accelerate PKU research has only just begun. The focus of the registry in its second year will be to continue to gather information from current participants as they age and reach out to those who have yet participated. To join the PKU registry go to https://pku.iamrare.org.

550 ENROLLED IN 41 STATES & 5 COUNTRIES
FUNDRAISING FOR A CURE

NPKUA had another successful year raising funds across the country under the Lifting the Limits for PKU campaign, which was designed to grow the NPKUA Fund substantially by hosting large fundraising events in targeted cities with the help of PKU families. The first event was in Buffalo, NY, raising awareness and $30,000 for PKU research. The second event was an opportunity presented because of the first-ever golf outing in 2016 in Virginia. In 2017, the NPKUA partnered with the Nicklaus Children’s Health Care Foundation as beneficiaries for the annual Croighton Farms Invitational hosted by Jack Nicklaus. With the event bringing in over $1 million dollars, the NPKUA was presented with a check for $500,000. The NPKUA looks forward to a continued partnership and also hosting key galas through Lifting the Limits for PKU in the years to come. These events are truly building the NPKUA Fund to help further new treatments and cure-based research. **Thank you to our families for their time and dedication to help us Lift the Limits for PKU!**

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MATERNAL PKU MENTOR PROGRAM

The Maternal PKU Mentor 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. Currently, the program has 10 mentors supporting 18 mentees, and to date, there have been 31 successful pregnancies. More information can be found at www.adultswithpku.org.

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ADVOCACY HILL VISIT

In May of 2017, NPKUA staff and volunteers joined forces on Capitol Hill to build critical support for the Medical Nutrition Equity Act (MNEA) in Congress. Advocacy efforts to pass the legislation by the end of 2018 are underway. Visit www.NPKUA.org to learn more about the bill and how you can help us ensure all individuals with PKU have access to the treatment they need.
Please join us for our next conference in Atlanta, GA, July 5-8, 2018. The conference will include exciting general sessions on the history and future of PKU, research, informational breakout sessions for parents, teens and adults, important networking time, and always fun. Here is your chance to meet researchers, industry professionals, and individuals and families with PKU. Register for early bird pricing by April 30, 2018. www.npkua.org/NewsEvents/Conferences