Dear Friends,

2018 marked the first decade of work for the National PKU Alliance. In 2008, several local groups across the country agreed to join forces to form the NPKUA believing that their collective efforts in combining resources, centralizing research grants and creating a national voice for PKU would truly change how PKU is experienced and how it will be cured.

I believe in that dream and so do you.

A decade ago, research for PKU was scattered and almost nonexistent. Since then, you helped us invest more than $3 million for PKU research. These funds led to new scientific knowledge on PKU, the acceleration of clinical trial programs, and leveraged much-needed federal research dollars for PKU.

A decade ago, there was no central source for scientifically based information on PKU. Your support has created a go-to place for new families, adults, and every stage in between, to get the information they need on how to best manage their PKU. The NPKUA also now hosts the largest gathering in the world of PKU adults, families, medical professionals, and industry supporters every two years.

A decade ago, there was no national voice for PKU. Now we are heard in the halls of Congress each year fighting for medical foods coverage, represented at the FDA during the clinical trials process and called upon by several federal agencies for our expertise in the unmet need of PKU.

Thank you for your tireless efforts, support and guidance in making our first decade a resounding success. However, our work is not done. I look forward to working with you in the next decade as we continue the fight for access to all treatments, support the most vulnerable in our community and work with industry partners for new treatments and a cure.

In gratitude,

Christine S. Brown
NPKUA Executive Director

ADVANCING THE DREAM IN 2018

The NPKUA held another record-breaking PKU conference in Atlanta, GA, July 5-8. It is the largest PKU gathering in the world with 665 attendees including families, researchers, and industry professionals from 34 states and 8 countries. The conference opened with the keynote presentation by Geir Ivar Følling Elgjo, grandson of the scientist who discovered PKU, Dr. Asbjørn Følling. Through various keynote presentations and educational breakouts, attendees were provided new information on PKU research advancements and future outlook, the importance of clinical trials, tips to better manage PKU, the PKU Patient Registry, advocacy, and much more. Two interactive panels were very well attended that focused on teens with PKU and research projects. The conference continues to create an avenue for families to network with each other and provide mutual support.

We challenged the attendees with a Text-to-Donate campaign and they exceeded our expectations by donating an additional $35,542 to NPKUA programs at the conference. Thank you!

The PKU Hero Award Banquet was filled with many accolades to share including a PKU Pioneer Award presented to Dr. Robert Guthrie, accepted by his daughter Patricia; recognition to Christineh Sarkissian and Ray Stevens for their instrumental work on Palynziq™; Biomarin with an Industry Champion Award in recognition of Palynziq™, accepted by Elaina Jurecki; and the PKU Hero Award presented to Dr. Barbara Burton, deserving of being recognized for her many years of service and dedication to PKU in her profession as a physician and beyond.

Thank you to our sponsors for supporting another important conference for PKU! Thank you to those volunteers that helped to make it a success!

ACCELERATING DRUG DEVELOPMENT

One of the biggest gaps in PKU drug development is the lack of regulatory agreement on what should be measured in the clinical trial process. As a national patient organization, the NPKUA is in a unique position to address this challenge for all drug developers. As a result, the NPKUA brought together officials from the FDA, industry, neuropsychologists and PKU clinicians to discuss this challenge in July. Participants developed a framework to better determine what meaningful, patient-centered and scientific endpoints should be considered and accepted in future clinical trials for adults. This meeting has resulted in supporting a multicenter study to validate these new potential endpoints.

Stay tuned to learn more about our progress!

COALITION ADVOCATES STRONG FOR RARE DISEASE

In June of 2018, more than 90 advocates (rare disease patients, family members, doctors and scientists) gathered on Capitol Hill for the first Patients & Providers for Medical Nutrition Equity (PPMNE) Lobby Day. Comprised of representatives from more than 30 patient organizations and hospitals, this coalition is formally taking up the fight our community has been waging for a long time: to obtain fair and equitable coverage for medical nutrition no matter where you live, what type of insurance you have, your diagnosis, age, gender or income. The advocates visited approximately 100 congressional offices and the coalition also hosted a Congressional Briefing for staff to hear first-hand from clinicians and patients about why medical nutrition is essential.

It was wonderful to see advocates from both the Inborn Errors of Metabolism (IEM) and Gastrointestinal (GI) disorder communities working together towards a common goal!

Join us for our next lobbying efforts May 6-7, 2019!

Thank you to those volunteers that helped to make it a success!
PKU PATIENT REGISTRY CONTINUES TO GROW

Thanks to the commitment of over 750 adults and children, the PKU Patient Registry remains as active as ever. Since 2017, registry participants have completed over 5,000 surveys containing important information about the natural history of PKU. The Registry has been used to recruit subjects for clinical trials, and registry data (illustrated in these graphs) was presented at scientific meetings around the world. New to the Registry this year was the addition of a validated PKU Quality of Life survey for adults and the opportunity for registry participants to access genetic testing at a reduced cost. Look for exciting updates in 2019 and monthly incentives for our active participants. Now is a great time to enroll by going to https://pku.iamrare.org.

LIFTING LIMITS IN BIG PLACES

NPKUA had a very successful year with fundraising by Lifting the Limits for PKU in two major cities and a famous golf tournament. The first event which elevated awareness for PKU and raised funds was held on May 5, in Denver, CO, bringing in over $400,000 for PKU research. The second event was another golf tournament in which NPKUA partnered with the Nicklaus Children’s Health Care Foundation (NCHCF) as beneficiaries for the annual Creighton Farms Invitational, hosted by Jack Nicklaus. The NPKUA was presented with a check for $505,000 from the NCHCF. In October, NPKUA held a gala in New York City, NY, with a record-breaking amount of over $600,000 raised. The NPKUA will hold another gala on April 27, 2019 in Philadelphia and will partner with the NCHCF for the Creighton Farms Invitational in August, 2019, and are anticipating 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. Please consider co-chairing an event if you live near a major city.

Thank you to our families for their time and dedication to help us Lift the Limits for PKU!

MATERNAL PKU MENTOR PROGRAM

Over the last six years the Maternal PKU Mentor Program has supported women with PKU who are pregnant or considering becoming pregnant by establishing an invaluable 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 17 mentees. To date, the Maternal PKU Mentor Program has supported 37 successful pregnancies. More information can be found at www.adultswithpku.org.

THEY SAY RAISING A CHILD TAKES A VILLAGE, WELL SO DOES PREGNANCY.

ESPECIALLY WHEN YOU HAVE PKU. THE MPKU PROGRAM AND MY MENTOR,

CARRIE, ARE A PART OF MY VILLAGE.

I DON’T KNOW HOW I WOULD HAVE GOTTEN THROUGH WITHOUT THEM.

OUR PASSION FOR THE NPKUA’S MISSION TO IMPROVE THE LIVES

OF THOSE WITH PKU, ESPECIALLY OUR KIDS, IS WHAT MOTIVATED US

TO HOST A LIFTING THE LIMITS EVENT. WE WERE DRIVEN TO RAISE

FUNDS FOR RESEARCH, AS WE ARE HOPEFUL TO SEE A CURE IN OUR

CHILDREN’S LIFETIMES. - MICHELLE AND EMELE, DENVER CO-CHAIRS
A few years ago, the NPKUA conducted a landmark study about the number of PKU patients being treated for their PKU. The results surprised us. Of the 16,500 Americans living with PKU, our study discovered that 8,000 of them had not been in a medical center for at least two years. This sobering fact led us to develop a Back to Care Coalition. Now, centralized messaging goes out to the entire community from all of our partners leading them back to a central hub at the NPKUA. Here, adults finding their way back to treatment can get connected to a specialized medical center, learn about resources, as well as get support in their PKU journey.