The National PKU Alliance (NPKUA) and NORD Launch Largest-Ever Study of Phenylketonuria (PKU)

Research study is open to participants worldwide to advance understanding and treatments for rare disease causing neurological problems impacting individual development, cognition, mood and concentration.

Tomahawk, WI, January 17, 2017— The National PKU Alliance (NPKUA) and the National Organization for Rare Disorders (NORD) today launched the largest-ever study to research phenylketonuria (PKU), an inherited metabolic disorder that, if untreated, can cause significant neurological problems. Even with treatment following diagnosis, individuals can experience a variety of neurological symptoms including cognitive impairment and problems with behavior and mood. PKU currently has no cure.

The new study, PKU Patient Registry, creates a platform for patients around the world to share information about PKU. Its purpose is to build an international resource to be used by scientists in future research.

"Participating in the PKU Patient Registry is the most critical thing that adults and parents can do to accelerate the development of new research and new therapies for PKU," announced Christine S. Brown, Executive Director, NPKUA.

To help drive awareness and participation, the NPKUA will be reaching out to the PKU community through social media, member organizations, clinicians and industry partners to learn about the registry.

"Our goal is to enroll as many patients, or their parents or legal guardians, as possible," said Brown. "The success of the registry is dependent upon community participation."

PKU Patient Registry is a natural history study that consists of electronic surveys to collect information about the patient experience and disease progression. Patients, or their caregivers or guardians, can enter information from anywhere in the world. The data is made anonymous and stored securely in an online portal called a registry. The NPKUA may share the data with individuals or institutions conducting research or clinical trials, as approved by the study's governing board that includes scientists, doctors and patient advocates.

The NPKUA is launching the study in collaboration with the NORD, an independent charity that built its natural history study platform as part of its mission to help identify and treat all 7,000 rare diseases. The NPKUA is a member of NORD and the organizations work together to eliminate the challenges that rare disease patients face.

"NORD's natural history studies platform empowers patients and families to drive research and eliminate some of the unknowns that still exist in rare diseases," said NORD President and CEO Peter L. Saltonstall. "We are glad to be working with The National PKU Alliance on this important project."

Phenylketonuria (PKU) is a rare, inherited metabolic disorder that occurs in approximately one out of every 15,000 people. PKU is characterized by the inability of the body to utilize phenylalanine (Phe) – an essential amino acid found in most protein. PKU is caused by a deficiency in the phenylalanine

hydroxylase (PAH) enzyme that converts Phe to tyrosine. Without a functional PAH enzyme, Phe accumulates in the blood, brain, and other body tissues. High levels of blood Phe are toxic to the central nervous system and can cause severe neurological complications and intellectual disabilities.

For more information, visit pku.iamrare.org.

About NPKUA

The National PKU Alliance mission is to improve the lives of individuals associated with PKU and pursue a cure. The NPKUA was formed in 2008 by parents, grandparents, and individuals across the country just like you. It's the first national non-profit organization to unite adults, families, regional and statewide PKU organizations, the medical community and PKU-friendly businesses to make a difference in the lives of people with PKU.

About National Organization for Rare Disorders (NORD)

The National Organization for Rare Disorders (NORD)® is the leading independent advocacy organization representing all patients and families affected by rare diseases. NORD is committed to the identification, treatment and cure of the 7,000 rare diseases that affect 30 million Americans, or 1 in every 10 people. NORD began as a small group of patient advocates that formed a coalition to unify and mobilize support to pass the Orphan Drug Act of 1983. For more than 30 years, NORD has led the way in voicing the needs of the rare disease community, driving supportive policies and education, advancing medical research, and providing patient and family services for those who need them most. NORD represents more than 250 disease-specific member organizations and their communities and collaborates with many other organizations in specific causes of importance to the rare disease patient community.

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