

The National PKU Alliance

Aiming to accelerate PKU research and work toward a cure, the National PKU Alliance has partnered with Baby Genes to create a program to allow easy access to genetic testing for individuals and their families with PKU.

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The National PKU Alliance (NPKUA) has partnered with Baby Genes Inc. to assist hundreds of people diagnosed with Phenylketonuria (PKU) obtain simple and affordable access to mutation analysis of the PAH gene responsible for PKU. PKU is an inherited metabolic disorder that is diagnosed at birth through newborn screening. Baby Genes' full gene sequencing technology provides accurate, efficient results identifying specific mutations associated with PKU. Test results will be made available in 96 hours after testing has started. Currently, access to mutation analysis is not standard of care in this rare disease.

There is no cure for PKU and lifelong treatment is difficult and expensive imposing unmanageable burdens for many individuals and their families. Through the Baby Genes program, individuals will learn their specific gene mutations which will facilitate the advancement of new treatments and provide a better understanding of the disease process for those in PKU research.

This program is currently open to any individual or family member enrolled in the PKU Patient Registry recently launched by the National PKU Alliance. The goal of the PKU Patient registry is to collect information to help medical providers better understand the natural history and progression of PKU and provide opportunity for more robust research and ultimately a cure.

About The National PKU Alliance

The National PKU Alliance's mission is to improve the lives of families and individuals associated with PKU through research, support, education and advocacy while ultimately seeking a cure. Formed in 2008, NPKUA is the first national non-profit to unite adults, families, regional and statewide PKU organizations, the medical community and PKU-friendly businesses to make a difference in the lives of individuals living with PKU. To learn more about the NPKUA and the PKU Patient Registry visit NPKUA.org.

About Baby Genes

Baby Genes Inc. is a CLIA-certified, diagnostic laboratory that offers advanced newborn testing using Next-Generation Sequencing (NGS). The Baby Genes Newborn Panel tests 106 genes for abnormalities that are linked to 171 hereditary disorders. The Baby Genes Newborn Test can be used as a confirmatory/reflex test for kids with abnormal NBS results, as a supplement to state-based newborn screening or as a family planning/carrier test. Baby Genes is located in Golden, Colorado and their website is www.BabyGenes.net.

To learn more about this opportunity for PAH gene sequencing please contact Eileen Blakely at eileen.blakely@npkua.org.