What is PKU?

PKU is short for phenylketonuria, also known as PAH deficiency, which is a rare genetic condition.

This condition disrupts neurotransmitters in the brain causing an imbalance in mood, learning, memory, and motivation.

PKU patients have strict diets since they have difficulty breaking down phenylalanine (Phe) commonly found in many foods, such as chicken, meat, eggs, dairy, nuts, grains, and legumes.

In the U.S.:

- About 16,500 people are living with PKU.
- About 350 babies are born with PKU each year.

There is a 1 in 4 chance of having a baby with PKU when both parents are carriers.

For over 15 years, BioMarin has been a pioneer in ongoing research to help improve the lives of PKU patients including:

- 3 therapies for PKU:
  - 2 approved treatments
  - 1 gene therapy under development

- 78 clinical studies on PKU:
  - 40 conducted internally by BioMarin
  - 38 externally sponsored by BioMarin

- 7,000 patients treated globally

- 106 publications on PKU:
  - 54 publications authored by BioMarin
  - 52 externally developed publications

Please go to PKU.com to learn more about this rare disease and access other resources available.