

PKU Selected Publications

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Pegvaliase / Palynzia	Harding CO, Whitehall KB, Lilienstein J, et al. Long-term management strategies for pegvaliase use in phenylketonuria: Lessons learned from the phase 3 PRISM open-label extension study. <i>Genet Med.</i> 2025;101459. DOI: 10.1016/j.gim.2025.101459
	Cooney E, Ammous Z, Bender T, et al. Lessons learned from 5 years of pegvaliase in US clinics: A case series. <i>Mol Genet Metab Rep.</i> 2025;42 DOI: 10.1016/j.ymgmr.2024.101181
	Burton BK, Clague GE, Harding CO, et al. Long-term comparative effectiveness of pegvaliase versus medical nutrition therapy with and without sapropterin in adults with phenylketonuria. <i>Mol Genet Metab.</i> 2024;141(1):108114. DOI: 10.1016/j.ymgme.2023.108114
	Harding CO, Longo N, Northrup H, et al. Pegvaliase for the treatment of phenylketonuria: Final results of a long-term phase 3 clinical trial program. <i>Mol Genet Metab. Rep.</i> 2024 Apr 23;39:101084. DOI: 10.1016/j.ymgmr.2024.101084
	Rohr F, Burton B, Dee A, et al. Evaluating change in diet with pegvaliase treatment in adults with phenylketonuria: Analysis of phase 3 clinical trial data. <i>Mol Genet Metab.</i> 2024;141(3):108122. DOI: 10.1016/j.ymgme.2023.108122
	Sigg MA, Wilson C, Clague GE, et al. Pegvaliase treatment normalizes blood neurotransmitter metabolites in adults with phenylketonuria. <i>Mol Genet Metab.</i> 2024;143(3) DOI: 10.1016/j.ymgme.2024.10858
Kuvan	Feillet F, Ficicioglu C, Lagler FB, et al. Efficacy and safety of sapropterin before and during pregnancy: Final analysis of the Kuvan® Adult Maternal Paediatric European Registry (KAMPER) maternal and Phenylketonuria Developmental Outcomes and Safety (PKUDOS) PKU-MOMs sub-registries. <i>J Inherit Metab. Dis.</i> 2024; 1-15. DOI: 10.1002/jimd.12724
Phenylketonuria	van Wegberg AMJ, MacDonald A, Ahring K, et al. European guidelines on diagnosis and treatment of phenylketonuria: First revision. <i>Mol Genet Metab.</i> 2025;145(2):109125. DOI: 10.1016/j.ymgme.2025.109125
	Smith WE, Berry SA, Bloom K, et al. Phenylalanine hydroxylase deficiency diagnosis and management: A 2023 evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG). <i>Genet Med.</i> 2025;27(1):101289. DOI: 10.1016/j.gim.2024.101289
	Burton BK, Hermida Á, Bélanger-Quintana A, et al. Management of early treated adolescents and young adults with phenylketonuria: Development of international consensus recommendations using a modified Delphi approach. <i>Mol Genet Metab.</i> 2022;137(1-2):114-126. DOI: 10.1016/j.ymgme.2022.07.012

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