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Selected Publications List Phenylketonuria

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Burden of Illness	Burton B, Hermida A, Belanger-Quintana A, et al. Management of early treated adolescents and young adults with phenylketonuria: Development of international consensus recommendations using a modified Delphi approach. *Molecular Genetics & Metabolism* 2022; 137: 114-126. DOI: 10.1016/j.ymgme.2022.07.012
	Jurecki ER, Cederbaum S, Kopesky J, et al. Adherence to clinic recommendations among patients with phenylketonuria in the United States. *Molecular Genetics & Metabolism* 2017; 120: 190-197. DOI: 10.1016/j.ymgme.2017.01.001
	Bilder DA, Noel JK, Baker ER, et al. Systematic review and meta-analysis of neuropsychiatric symptoms and executive functioning in adults with phenylketonuria. *Developmental Neuropsychology* 2016; 41: 245-260. DOI: 10.1080/87565641.2016.1243109
Sapropterin dihydrochloride	Muntau AC, Adams DJ, Belanger-Quintana A, et al. International best practice for the evaluation of responsiveness to sapropterin dihydrochloride in patients with phenylketonuria. *Molecular Genetics & Metabolism* 2019; 127: 1-11. DOI: 10.1016/j.ymgme.2019.04.004
	Waisbren S, Burton BK, Feigenbaum A, et al. Long-term preservation of intellectual functioning in sapropterin-treated infants and young children with phenylketonuria: A seven-year analysis. *Molecular Genetics & Metabolism* 2021; 132 (2): 119-127. DOI: 10.1016/j.ymgme.2021.01.001
	Longo N, Arnold GL, Pridjian G, et al. Long-term safety and efficacy of sapropterin: The PKUDOS registry experience. *Molecular Genetics & Metabolism* 2015; 114 (4): 557-563. DOI: 10.1016/j.ymgme.2015.02.003
Pegvaliase	Longo N, Dimmock D, Levy H, et al. Evidence- and consensus-based recommendations for the use of pegvaliase in adults with phenylketonuria. *Genetics in Medicine* 2019; 21: 1851-1867. DOI: 10.1038/s41436-018-0403-z
	Lah M, Cook K, Gomes DA, et al. Real-world treatment, dosing, and discontinuation patterns among patients treated with pegvaliase for phenylketonuria: Evidence from dispensing data. *Molecular Genetics & Metabolism Reports* 2022; 33: 100918. DOI: 10.1016/j.ymgmr.2022.100918
	Adams D, Andersson HC, Bausell H, et al. Use of pegvaliase in the management of phenylketonuria: Case series of early experience in US clinics. *Molecular Genetics & Metabolism Reports* 2021; 28: 100790. DOI: 10.1016/j.ymgmr.2021.100790
	Thomas J, Levy H, Amato S, et al. Pegvaliase for the treatment of phenylketonuria: Results of a long-term phase 3 clinical trial program (PRISM). *Molecular Genetics & Metabolism* 2018; 124 (1): 27-38. DOI: 10.1016/j.ymgme.2018.03.006
	Harding C, Amato RS, Stuy M, et al. Pegvaliase for the treatment of phenylketonuria: A pivotal, double-blind randomized discontinuation Phase 3 clinical trial. *Molecular Genetics & Metabolism* 2018; 124 (1): 20-26. DOI: 10.1016/j.ymgme.2018.03.003

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