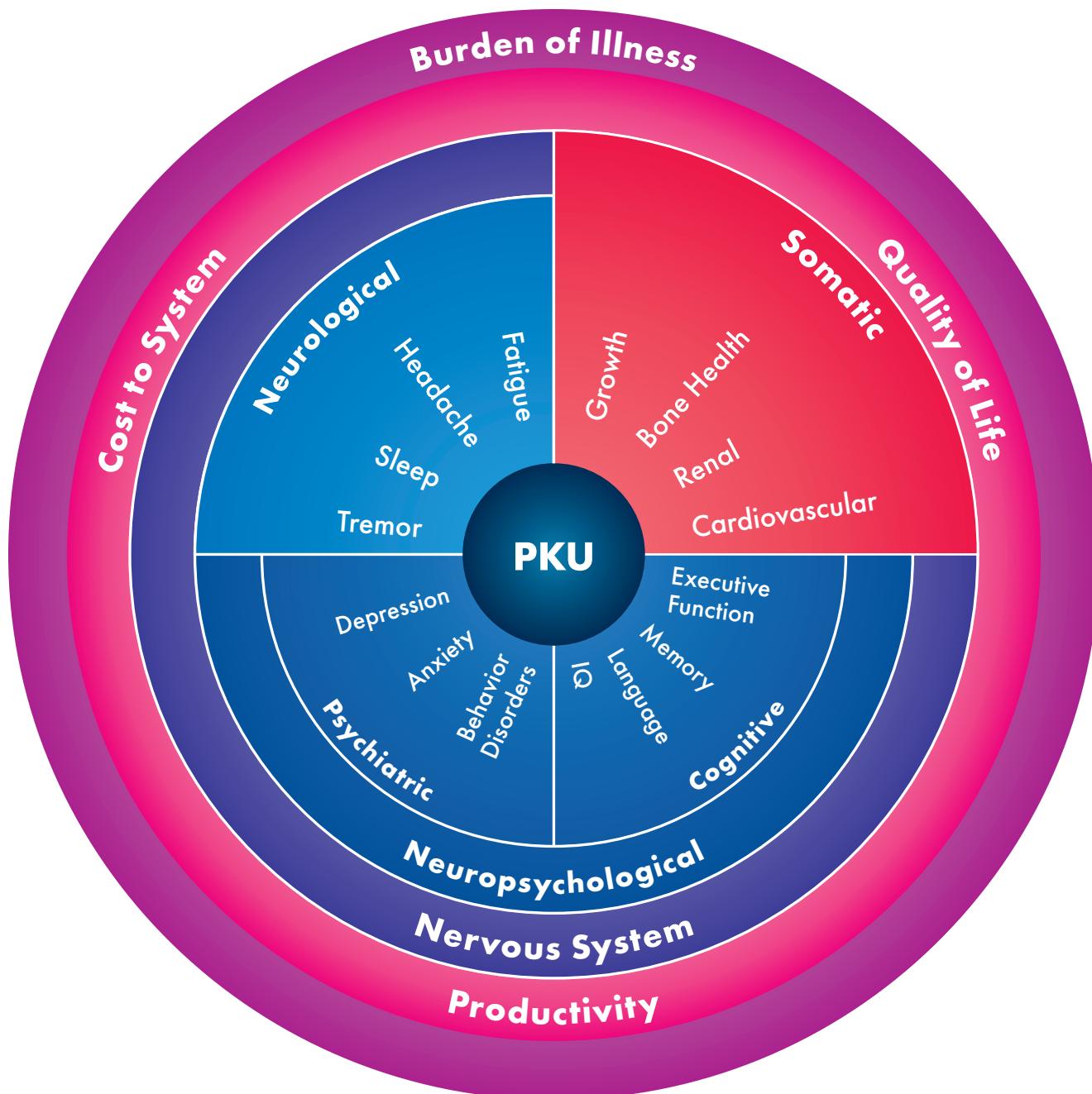


NEUROCOGNITIVE, NEUROLOGICAL, AND NEUROPSYCHOLOGICAL MANIFESTATIONS OF PHENYLCETONURIA¹⁻³

Phenylketonuria results in an accumulation of phenylalanine. Without lifelong treatment, elevated phenylalanine can cause harmful complications to the growth and function of the brain and body and affect the way a person thinks, feels and acts.



References

1. 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). *Genet Med.* 2025;27(1):101289.
2. Enns GM, Koch R, Brumm V, et al. Suboptimal outcomes in patients with PKU treated early with diet alone: Revisiting the evidence. *Mol Genet Metab.* 2010; 101(2-3): 99-109.
3. Ashe K, Kelso W, Farrand S, et al. Psychiatric and cognitive aspects of phenylketonuria: The limitations of diet and promise of new treatments. *Front Psychiatry.* 2019; 10: 561.