Phenylketonuria and BH4 Deficiencies

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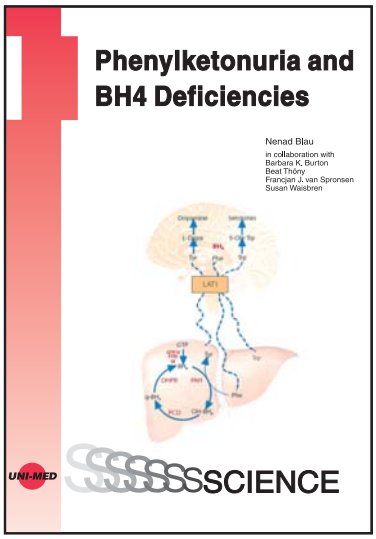

Thanks to newborn screening and early dietary therapy, individuals with PKU no longer experience intellectual disability (mental retardation). Nevertheless, many do not achieve their full potential. The establishment of uniform guidelines and improved management for PKU can lead to optimal outcomes in this metabolic disorder.
Two new approaches are currently available for the treatment of PKU in some patients:
- The use of tetrahydrobiopterin (BH4; sapropterin) in individuals with PKU with certain mutations of the phenylalanine hydroxylase gene.
- Supplementation with large neutral amino acids (LNAA) other than phenylalanine.
Further approaches, such as recombinant phenylalanine ammonia lyase (PAL), which degrades phenylalanine, and gene therapy, using viral or non-viral vectors to correct murine PKU, are at the experimental stage.
In this textbook past, present, and future efforts related to PKU and BH4 deficiencies are discussed. The reviews and scientific contributions in this book provide professionals, the patients and their families to understand PKU within a biochemical, neurological and psychosocial context.

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