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Advances of Phenylketonuria in Children

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Message from the Guest Editors

From1934 first detected phenyl pyruvic acid to 1963 a method for measuring phenylalanine from dried blood spots of newborns, it took almost 30 years. However, important drawbacks remain in the management of phenylketonuria (PKU) now. Adherence to dietary therapy, especially after the first decade of life, is often unsatisfactory, with consequences for neurocognitive function. Behavioral and emotional problems are still described in many continuously treated children and adolescents. The neuropathology of PKU remains a major knowledge gap. Sapropterin, an oral form of tetrahydrobiopterin, is an alternative pharmacological treatment for a subset of patients with PKU, mainly those with mild or moderate metabolic phenotypes. It is not known whether pegylated phenylalanine ammonia lyase, which requires daily subcutaneous injections, is safe and effective in children.

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