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Editorial Letter to Special Issue on Phenylketonuria

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More than 80 years ago, Asbjørn Fölling described a clinical case of children with severe mental retardation who excreted large amounts of phenylpyruvic acid in their urine, secondary to an increase of phenylalanine (PHE) in blood, calling it *idiocia fenilpiruvica*, which was later called phenylketonuria (PKU).[1]

Phenylketonuria (MIM # 261600) is an autosomal recessive human genetic disease caused by mutations in the gene that encodes the liver protein phenylalanine hydroxylase (EC 1.14.16.1).[2]

In 1961, Dr Robert Guthrie first described a method for determining PHE in dried blood collected on filter paper, allowing neonatal detection. This advance was the start of eliminating PKU as a cause of mental retardation in countries that have implemented screening programs.[3] Through these programs, the overall incidence of PKU is 1:10 000 newborns, with variation by ethnic group and existing consanguinity.[3]

From the beginning of neonatal diagnosis, the restriction of PHE is used as a treatment for PKU, preventing the neurological sequela that this disease causes. Optimal treatment requires diagnosis before 1 month of life, strict monitoring, and maintaining blood PHE levels between 2 and 6 mg/dL (120-360 µmol/L) throughout life.[4]

Scientific and technological advances in the last 15 years in the area of inborn errors of metabolism (IEM), especially concerning the development of innovative platforms of neonatal diagnosis and the discovery of new treatment and prevention alternatives, have led to changes in the traditional approach for health care.[5]

In Latin America, there is concern over the ever-increasing gap between scientific advances in developed versus developing economies. Developing economies are the norm in the region, which is reflected in the IEM neonatal screening programs.

In the last decade, some Latin American countries have had significant growth in the area of neonatal screening for PKU, and it is likely that several new screening programs will emerge in the coming years. This will significantly increase the number of individuals diagnosed with PKU and will enhance interdisciplinary groups dedicated to the monitoring and treatment of this disease.[6]

Based on these facts, the Latin American Society of Inborn Errors of Metabolism and Neonatal Research formed a committee of experts in PKU to create a protocol for the diagnosis and treatment of PKU and other hyperphenylalaninemias (HPAs) for Latin America. The goal was to unify criteria in neonatal screening, diagnosis, classification, treatment, and long-term monitoring and provide knowledge on new treatment alternatives for PKU.

The main objective of this supplement that focuses on PKU is for professionals involved in the care of individuals with HPA/PKU in Latin America to describe how they perform diagnosis and follow-up, determine which variables are important to consider, and how to apply them in their workplaces in order to evaluate their results.

This supplement provides an opportunity for discussion related to PKU and, in the near future, will allow for the formation of the first guidelines for the diagnosis and treatment for people with PKU/HPA in Latin America, which will improve the quality of life of people with PKU/HPA in the region.

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