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Title | Current practices and challenges in the diagnosis and management of PKU in Latin America: results from a multicenter survey
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The aim was to describe the current practices on diagnosis and dietary management of phenylketonuria (PKU) in Latin America.

Methods: An online survey containing 44 questions regarding diagnosis and management of PKU was developed by a team of experts, and was targeted for health care professionals following PKU patients. After a pilot study, the final version was sent to 14 countries where Inborn Errors of Metabolism practitioners were found.

Results: Twenty-three centers from 13 countries were included. Respondents were mostly females (91%), had ≥45 years-old (61%) and worked with PKU for over 10 years (70%). Most countries (12/13) perform newborn screening for PKU. Most centers (15/23) started dietary treatment with phenylalanine (Phe) levels ≥360 µmol/l. Phe target throughout life was very heterogeneous among centers, with greater consistency at the age of 0-1 year (14/23 aimed Phe 120-240 µmol/l), and the lowest agreement >12 years (11 different targets reported). Regarding dietary resources, most countries (10/13) had available only unflavored powder amino acids substitute, which was fully provided by the government. Low-protein foods for PKU were not available in 8/13 countries; and even when available, they were not affordable. Only 3/13 countries had an adequate regional database of Phe content of foods, and only 4/23 centers had a nutrient analysis software program. The obstacles for treatment most commonly perceived were: lack of financial resources (62%), availability of low-protein foods (60%) poor adherence and lack of technical resources to manage the diet (50% each).

Discussion: we found a very heterogeneous scenario in the dietary management of PKU in Latin America, indicating a major need for regional guidelines. In common, most countries experience a lack of resources for both patients and health care professionals.

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