Current practices and challenges in the diagnosis and management of PKU in Latin America: a multicenter survey

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Latin America comprises 20 countries with an ethnically diverse population of over 650 million. With a complex political and economic background, these countries face many challenges in the care of patients with rare diseases.

In order to describe the current practices on diagnosis and dietary management of phenylketonuria (PKU) in Latin America, we performed an online survey targeted for health care professionals following PKU patients.

Subjects
Respondents were mostly females (91%), had ≥45 years-old (61%) and worked with PKU for over 10 years (70%).

Phe and Tyr monitoring
• High heterogeneity in blood monitoring
• The highest agreement (65%) was for Phe measurement once a week or more in infants <1 year
• 20% rarely or never measured tyrosine

Dietary resources
• Only 3 countries had a suitable database of Phe content of foods
• Only 4 centers had a nutrient analysis software program
• 10 countries had available only unflavored powder amino acids substitute (fully provided by the government)
• Low-protein foods were not available in 8/13 countries

Alternative therapies
• BH4 was approved in 7 countries, LNAA in 2 and GMP in 1

Barriers to treatment
• The obstacles for treatment most commonly perceived were: availability of low-protein foods (62%), lack of financial resources (60%), poor adherence and lack of technical resources to manage the diet (50% each)

NBS
12/13 countries performed newborn screening for PKU

Breastfeeding
All except 2 centers recommended partial breastfeeding maintenance

Initiation of treatment
Most centers (65%) started dietary treatment with phenylalanine (Phe) levels ≥360 μmol/l.

Figure 1 shows Phe targets throughout life in the centers studied, and total protein prescriptions are described in Figure 2.

Figure 2: Total protein (natural + protein substitute) prescriptions according age groups in the centers studied (n=23).

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, which may be impairing treatment outcomes.