

Article title: Diagnostic and management practices for phenylketonuria in 19 countries of the South and Eastern European Region: survey results

Journal: European Journal of Pediatrics

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Online resource 3

Summary of main challenges in relation to screening, confirmatory diagnosis and treatment of PKU, and areas for improvement in PKU care (N=31)

<i>Main challenges in screening, confirmatory diagnosis and treatment of PKU^a</i>	<i>Responses % (n)</i>	<i>Areas for improvement in PKU care^a</i>	<i>Responses % (n)</i>
Lack of BH4-responsiveness testing/treatment (in all age groups)	12.9% (4)	Reimbursement for low-protein foods	16.1% (5)
Lack of funds for PKU treatment and management	9.7% (3)	Broader range of Phe-free protein substitutes/low-protein foods	16.1% (5)
Lack of TMS for diagnosis	6.5% (2)	Adequate funds for PKU treatment and management	16.1% (5)
Lack of skilled/coordinated treatment team	6.5% (2)	BH4 loading tests/treatment	12.9% (4)
		Setting up of dedicated specialist team.	12.9% (4)
		Provision of dietician	12.9% (4)
		Improved diagnostic testing (more methods/improved application)	9.7% (3)
		Newborn screening programme (Establishment/increase budget)	9.7% (3)
		Closer collaboration between medical service and external companies/organisations (pharmacies/PKU associations)	9.7% (3)
		Improved social support for patients and their families	6.5% (2)
Other ^b	25.8% (8)	Other ^c	3.2% (1)
No answer	48.4% (15)	No answer	41.9% (13)

^aOpen answers provided by participants were summarised and grouped and only those challenges/areas for improvement stated in more than 2 questionnaires are included as separate row headers in the table; ^b'Other' included: 'lack of treatment options beyond low-Phe diet', 'lack of all methods for confirmatory diagnosis', 'lack of dietician', 'lack of newborn screening', 'lack of PKU product availability', 'lack of reimbursement for low-protein foods', 'high cost of low-protein foods', 'lack of education'; ^c'other' included: 'Separate care for adult and paediatric patients with PKU' BH4, sapropterin dihydrochloride; Phe, phenylalanine; PKU, phenylketonuria; TMS, tandem mass spectroscopy