Molecular Genetics and Metabolism 118 (2016) 72–83

Contents lists available at ScienceDirect

Molecular Genetics and Metabolism

journal homepage: www.elsevier.com/locate/ymgme

Minireview

Updated, web-based nutrition management guideline for PKU: An evidence and consensus based approach

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ARTICLE INFO

Article history:
Received 20 January 2016
Received in revised form 15 April 2016
Accepted 15 April 2016
Available online 17 April 2016

Keywords:
Phenylketonuria
PKU
Phenylalanine
Guidelines
Recommendations
Inherited metabolic disorders

ABSTRACT

Background: In 2014, recommendations for the nutrition management of phenylalanine hydroxylase deficiency were published as a companion to the concurrently published American College of Medical Genetics and Genomics guideline for the medical treatment of phenylketonuria (PKU). These were developed primarily from a summary of findings from the PKU scientific review conference sponsored by the National Institutes of Health and Agency for Healthcare Research & Quality along with additional systematic literature review. Since that time, the Genetic Metabolic Dietitians International and the Southeast Regional Newborn Screening and Genetics Collaborative have partnered to create a web-based technology platform for the update and development of nutrition management guidelines for inherited metabolic disorders.

Objective: The purpose of this PKU guideline is to establish harmonization in treatment and monitoring, to guide the integration of nutrition therapy in the medical management of PKU, and to improve outcomes (nutritional, cognitive, and developmental) for individuals with PKU in all life stages while reducing associated medical, educational, and social costs.

Methods: Six research questions critical to PKU nutrition management were formulated to support guideline development: Review, critical appraisal, and abstraction of peer-reviewed studies and unpublished practice literature, along with expert Delphi survey feedback, nominal group process, and external review from metabolic physicians and dietitians were utilized for development of recommendations relevant to each question. Recommendations address nutrient intake, including updated protein requirements, optimal blood phenylalanine concentrations, nutrition interventions, monitoring parameters specific to life stages, adjunct therapies, and pregnancy and lactation. Recommendations were graded using a rigorous system derived from the Academy of Nutrition and Dietetics.

Results and Conclusion: These guidelines, updated utilizing a thorough and systematic approach to literature analysis and national consensus process, are now easily accessible to the global community via the newly developed digital platform. For additional details on specific topics, readers are encouraged to review materials on the online portal: https://GMDI.org/.

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Abbreviations: ACMG, American College of Medical Genetics; AGREE II, Appraisal of Guidelines for Research and Evaluation; BH4, tetrahydrobiopterin; BMD, bone mineral disease; DRI, dietary reference intake; DXA, dual x-ray absorptiometry; EF, executive function; FFM, fat-free mass; GMDI, Genetic Metabolic Dietitians International; IMD, inherited metabolic disorder; LNAA, large neutral amino acid; MeSH, medical subject heading; MPKU, maternal PKU syndrome; MRI, magnetic resonance imaging; MSUD, Maple Syrup Urine Disease; NIH, National Institutes of Health; PAH, phenylalanine hydroxylase; PHE, phenylalanine; PI, principal investigator; PICO, population, intervention, comparison, and outcomes; PKU, phenylketonuria; QoL, quality of life; SERC, Southeast Regional Newborn Screening and Genetics Collaborative; TYR, tyrosine.

http://dx.doi.org/10.1016/j.ymgme.2016.04.008
1096-7192/© 2016 Published by Elsevier Inc.

Contents

1. Introduction

Phenylketonuria (PKU), an autosomal recessive inherited metabolic disorder (IMD), is characterized by abnormally high concentrations of blood phenylalanine (PHE) and production of phenylketones resulting from impaired phenylalanine hydroxylase (PAH) function. Also referred to as PAH deficiency, the disorder represents a continuum of impairment in enzyme function. Failure or delay in treatment can result in irreparable neurologic damage and severe developmental delay [1,2]. The main goal of PKU therapy is to maintain blood PHE concentrations within a recommended treatment range of 120–360 μmol/L and to support nutritional needs so that growth and development are within the normal range [2]. This is accomplished by restriction of dietary PHE to that required for anabolism, consumption of medical food products to ensure optimal nutrient intake, and, in some individuals, adjunct pharmacotherapy [1–3].

Widespread consensus exists regarding the importance of blood PHE control and dietary treatment [2,3]. In response to the 2013 recommendations by the National Institutes of Health (NIH) and Agency for Healthcare Research & Quality (AHRQ), the American College of Medical Genetics and Genomics (ACMG) guideline for diagnosis and medical management of PKU (ACMG Guideline) was published in 2014 in conjunction with the Genetic Metabolic Dietitians International (GMDI) and Southeast Regional Newborn Screening and Genetics Collaborative (SERC) evidence- and consensus-based recommendations for nutrition management of PAH deficiency (GMDI/SERC Recommendations) [2,3]. This guideline advances the 2014 GMDI/SERC recommendations for nutrition management of PAH deficiency by incorporating a rigorous and expanded review of the latest research, grading the body of evidence, and utilizing a web-based technology that supports global open access. This process also resulted in revised recommendations for protein requirements.

Development of this guideline is part of a multi-year project undertaken by GMDI and SERC to develop nutrition management guidelines for rare IMDs to foster optimum nutrition management of affected individuals, to reduce the uncertainty and variability in management, and to direct future research. This is the second IMD guideline published by the partnership, building on the experience of the nutrition management guideline developed for Maple Syrup Urine Disease (MSUD) [4]. The

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