Phenylketonuria Scientific Review Conference: State of the Science and Future Research Needs

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Background

Phenylketonuria (PKU) is an amino acidopathy typically caused by mutations in the PAH gene. Newborn screening for PKU continues to be a successful public health program. Early identification and initiation of dietary treatment in infants with PKU prevents intellectual disabilities. The National Institutes of Health (NIH) published a statement on screening and management for PKU as the result of a Consensus Conference held in 2000.[1]

A new pharmacologic intervention, tetrahydrobiopterin (BH4), can allow diet liberalization in those who respond. The combination of new treatments, screening technologies, and more literature on outcomes for persons with PKU warrant review of those guidelines to determine their current applicability.

Methods

An NIH Scientific Review Conference will be held February 22-23, 2012 in Bethesda, MD to consider recent research findings, current treatments, the role of BH4, and future research needs. Five working groups of topical experts (clinicians and researchers), patients and advocacy group members, and federal stakeholders have been established to answer specific questions related to the aims of the conference.

Related Activity

In a parallel and collaborative effort, an Evidence-based Practice Center (EPC) of the Agency for Healthcare Research and Quality[2] is conducting an evidence-based review[3] of the comparative effectiveness of BH4 and diet in the treatment of PKU. The EPC draft report is now available for comment until Oct. 18, 2011 at: http://effectivehealthcare.ahrq.gov/

Working Groups and Overarching Questions

Working Group 1: Diet Control and Management
Should the dietary recommendations that emerged from the 2000 Consensus Statement be changed? If so, what current knowledge would inform development of new recommendations?

Working Group 2: Pharmacologic Interventions
What is the role of BH4 in individuals with PKU?

Working Group 3: PKU and Pregnancy
What are the current dietary recommendations for women with PKU who become pregnant, and should they be modified?

Working Group 4: Long Term Outcomes and Follow-up
What should be the current recommendations for individuals with PKU across the lifespan in terms of diet and management(excluding pregnancy)?

Working Group 5: Molecular Testing, New Technologies, and Epidemiologic Considerations
Should there be any changes to the 2000 Consensus Statement regarding newborn screening and molecular testing for PKU?

Interested in attending the conference? Contact Melissa Parisi at: parisima@mail.nih.gov

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Working Group Structure

Impact of the PKU Conference and EPC Report

Together, the PKU Scientific Conference and the Evidence-based Practice Center report will provide evidence on important issues, recommend future research, and provide guidance to patients, their families, and health care professionals.

References and Resources

1. National Institutes of Health Consensus Development Panel. National Institutes of Health Consensus Development Conference Statement: Phenylketonuria: Screening and Management, October 16-18, 2000. *Pediatrics*. 2001;108(4):972-982.

2. Agency for Healthcare Research and Services. Effective Healthcare Program. See http://www.effectivehealthcare.ahrq.gov/.

3. Agency for Healthcare Research and Services. Methods Guide for Effectiveness and Comparative Effectiveness Reviews. See http://www.effectivehealthcare.ahrq.gov/index.cfm/search-for-guides-reviews-and-reports/?pageaction=displayproduct&productID=318.

A similar abstract was previously presented at the SSIEM Annual Meeting held February 27 - March 3, 2011