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 treatment of infants with PKU prevent intellectual disabilities. The National Institutes of Health (NIH) published a statement on screening and management of PKU as the result of a Consensus Conference held in 2000.

A new pharmacological intervention, tetrahydrobiopterin (BH4), can allow dietary treatment in those who respond. The combination of new treatments, screening technologies, and more active follow-up on outcomes of persons with PKU was anticipated to determine their applicability to existing guidelines.

**Background**

An NIH Scientific Review Conference will be held on June 22-23, 2012 in Bethesda, MD to assess recent research findings, current treatments, the use of BH4, and future research needs. Five working groups of top scientific, clinical, and investigational partners have been established to answer specific questions related to the aims of the conference.

**Methods**

An NIH Scientific Review Conference will be held on June 22-23, 2012 in Bethesda, MD to assess recent research findings, current treatments, the use of BH4, and future research needs. The national institutes of health Co-senior Development Panel on PKU and co-senior development conference statement on PKU screening and management in october 16-28, 2000. ed at i s 20 1.1.8(4) 72-982

**Working Groups and Overarching Questions**

**Working Group 1: Diet Control and Management**

Shou d the detailed recommendations that emerged from the 2000 Consensus Statement be changed? In o what current knowledge would o m development of new recommendations?

**Working Group 2: Pharmacological 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 Epidemiological Considerations**

Should there be any changes to the 2000 Consensus Statement regarding newborn screening and molecular testing for PKU?

**Related Activity**

In a parallel and collaborative effort, the Agency for Healthcare Research and Quality (AHRQ) is conducting an evidence-based review of the comparative effectiveness reviews (CER) for PKU. The AHRQ data and tools now available are comment until Oct 18 2011 at http:// effect vehealthca e.hsgov.

Inte rested? Attend the conference? Contact Melissa Pas at pas ma@ma l.n h.gov.

**References and Resources**


