



Effective Health Care

Pharmacologic Therapy versus Medical Foods for the Treatment of Phenylketonuria (PKU)

Nomination Summary Document

Results of Topic Selection Process & Next Steps

- Pharmacologic therapy versus medical foods and nondietary approaches for the treatment of phenylketonuria (PKU) will go forward for refinement as a systematic review. The scope of this topic, including populations, interventions, comparators, and outcomes, will be further developed in the refinement phase.
- When key questions have been drafted, they will be posted on the AHRQ Web site and open for public comment. To sign up for notification when this and other Effective Health Care (EHC) Program topics are posted for public comment, please go to <http://effectivehealthcare.ahrq.gov/index.cfm/join-the-email-list1/>.

Topic Description

Nominator: Organization

Nomination Summary: The nominator is interested in the comparative effectiveness of treating PKU with pharmacologic therapy versus medical foods. They are also interested in the comparative cost effectiveness of these treatments. The nominator states that pharmaceuticals for this condition are much more expensive than medical foods and have side effects that medical foods do not.

Population: All patients with PKU ages 0 years and up, including pregnant women

Intervention: Pharmacologic therapy (Sapropterin dihydrochloride, Kuvan)

Comparators: Phenylalanine restricted diet including the use of medical foods, amino acids, and micronutrients supplement

Outcomes: Cost, adverse effects, and improved health outcomes

Key Questions from Nominator: None

Considerations

- The topic meets all EHC Program selection criteria. (For more information, see <http://effectivehealthcare.ahrq.gov/index.cfm/submit-a-suggestion-for-research/how-are-research-topics-chosen/>.)

- PKU is a metabolic disorder caused by a genetic mutation that usually results from a deficiency of a liver enzyme involved in the breakdown of the essential amino acid phenylalanine, leading to a buildup of phenylalanine in the blood and tissues. Accumulation of phenylalanine in untreated PKU causes CNS toxicity, most notably mental deficiency with severe learning disabilities; organ damage; unusual posture; and can, in cases of maternal PKU, severely compromise pregnancy.
- Historically, the treatment of PKU has relied on nonpharmacologic therapies, with affected individuals instructed to follow a strict diet that is often difficult for patients to comply with. A new pharmacologic agent is now available and may benefit a subset of patients with PKU.
- This topic will move forward as a systematic review with a focus on pharmacologic therapy for PKU. Comparators will include specialized/restricted diets, including the use of medical foods, amino acids, and micronutrients. This review may also address the diagnostic accuracy and categorization of the disease.