

# Phenylketonuria (PKU)

## Condition Description

Phenylketonuria (PKU) is a disorder of amino acid metabolism whereby phenylalanine builds up in the blood to toxic levels causing severe intellectual disabilities, neurological symptoms, abnormal gait, and seizures soon after birth. Effects are not reversible with treatment.

## Range of Outcomes

With newborn screening most infants are found pre-symptomatically and treated immediately with a significantly restricted protein diet of special formula/ food. Even with treatment compliance, over half the children have a consistent pattern of deficiencies in executive function; including high incidence of ADHD, slower mental processing, poor reasoning and problem solving, perceptual motor problems, learning disability. (Waisbren) Behavioral and mental health problems may be noted. (Smith and Knowles) Allowing more time for learning and repetition may be needed as well as appropriate interventions if needed for behavior.

## Common Complications

Untreated children with persistent severe hyperphenylalaninemia (i.e., PKU) show impaired brain development. Signs and symptoms include microcephaly, epilepsy, severe intellectual disabilities, and behavior problems. The excretion of excessive phenylalanine and its metabolites can create a musty body odor and skin conditions such as eczema.

## Side Effects of Treatment

Balancing dietary levels of phenylalanine needed to promote healthy brain growth without allowing levels to go too high thus

causing harm is a challenge. The diet is quite restrictive and difficult for families to implement fully. Team assessment, monitoring and intervention related to needed services for neurological, behavioral, social and cognitive delays is indicated. Accommodations will always be needed for extremely restricted diet.

## For More Information

**This fact sheet was developed for use in determining eligibility for early intervention services only. For more complete information, the following resources might be useful:**

### MedlinePlus

MedlinePlus brings together authoritative information from NLM, the National Institutes of Health (NIH), and other government agencies and health-related organizations. Preformulated MEDLINE searches also give access to medical journals.

<http://medlineplus.gov/>

### Agency for Healthcare Research and Quality

Under its Evidence-based Practice Program, the Agency for Healthcare Research and Quality (AHRQ) is developing scientific information for other agencies and organizations on which to base clinical guidelines, performance measures, and other quality improvement tools. Contractor institutions review all relevant scientific literature on assigned clinical care topics and produce evidence reports and technology assessments, conduct research on methodologies and the effectiveness of their implementation, and participate in technical assistance activities.

<http://www.ahrq.gov/clinic/epcsums/lbwdisum.htm>



Minnesota Children with Special Health Needs (MCSHN)  
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[www.health.state.mn.us/mcshn](http://www.health.state.mn.us/mcshn)

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### E-Medicine

eMedicine's clinical knowledge base contains peer reviewed articles on a number of different health conditions. There is a pediatric section available.

<http://www.emedicine.com/>

### References

Smith and Knowles. "Behavior in early treated phenylketonuria: a systematic review.2000.

Welsh, "A prefrontal dysfunction model of early treated phenylketonuria", (1996).

Waisbren et al, "Review of neuropsychological functioning in treated phenylketonuria: an information processing approach", (1994).

Macdonald "Diet and compliance in phenylketonuria".(2000).

Diamond,"Phenylalanine levels of 6-10mg?dl may not be as benign as once thought".(1994)

