

Adult Genetic Disease ACT Sheet

Adult Phenylketonuria (PKU)

Condition Description: In Phenylketonuria (PKU) the phenylalanine from ingested protein cannot be metabolized to tyrosine because of deficient liver phenylalanine hydroxylase (PAH). This causes elevated phenylalanine. In North America, 200 patients with PKU enter adulthood each year. Adults with PKU not adherent to dietary phenylalanine restrictions, **particularly women considering pregnancy**, may be at risk for medical problems.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Confirm diagnosis by prior records or blood phenylalanine levels.
- Determine treatment history and current treatment status.
- Consult with PKU specialists (physician and dietician) to review therapeutic options and ensure care coordination.
- Assess school and work history.
- Consider neuropsychological testing.
- Explain to affected women the reproductive risks to their fetus.

Clinical Considerations: Confirm diagnosis by prior records or blood phenylalanine levels. Unless treated from infancy, PKU will cause irreversible mental retardation, hyperactivity, autistic-like features, and seizures. Treatment with a phenylalanine restricted diet usually prevents these symptoms. Dietitians experienced in the care of PKU are essential. Many affected individuals may have relaxed or discontinued their dietary restrictions which may lead to psychological, cognitive, behavioral, and emotional difficulties. Reasons for poor treatment adherence (e.g., taste, cost) should be evaluated. Detailed educational and work history may reveal associated functional problems. Coordination of special services (e.g., social services, vocational rehabilitation, payment for medical foods, preconceptional counseling, insurance, and medical care) may be required for appropriate care. PKU is not causally related to commonly encountered diseases of adults.

Maternal PKU requires **rigorous** dietary restrictions before and throughout pregnancy to protect the fetus from serious birth defects.

Additional Information:

[Medline Plus](#)

[New England Metabolic Consortium](#)

[Gene Tests/Gene Clinics](#)

Referral (local, state, regional and national):

[Testing](#)

[Clinical Services](#)

[Find Genetic Services](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site *(insert state newborn screening program website information)*

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

Local Resource Site *(insert local and regional newborn screening website information)*

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

APPENDIX: Resources with Full URL Addresses

Additional Information:

Medline Plus

<https://medlineplus.gov/genetics/condition/phenylketonuria/>

New England Metabolic Consortium

<https://www.newenglandconsortium.org/printable-transition-toolkits>

Referral (local, state, regional and national):

Testing

<http://www.ucsdglab.org/>

Clinical Services

<http://www.ncbi.nlm.nih.gov/sites/genetests/clinic?db=genetests>

Find Genetic Services

<https://clinics.acmg.net/>

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