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:

Genetics Home Reference
New England Metabolic Consortium
Gene Tests/Gene Clinics

Referral (local, state, regional and national):

Testing
Clinical Services
Find Genetic Services

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

LOCAL RESOURCES: Insert State newborn screening program web site links

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

Name	
URL	
Comments	

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

Name	
URL	
Comments	

APPENDIX: Resources with Full URL Addresses

Additional Information:

Genetics Home Reference
http://ghr.nlm.nih.gov/condition=phenylketonuria

New England Metabolic Consortium
http://www.newenglandconsortium.org/toolkit/transition.html

Referral (local, state, regional and national):

Testing
http://biochemgen.ucsd.edu/UCSDW3BG/LabChoose.asp

Clinical Services
http://www.ncbi.nlm.nih.gov/sites/genetests/clinic?db=genetests

Find Genetic Services
http://www.acmg.net/GIS/Disclaimer.aspx

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®