

## ***Phenylketonuria (PKU) for Healthcare Providers***

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools ([www.gemssforschools.org](http://www.gemssforschools.org)).

### Physical characteristics and/or symptoms

Children with PKU do not have any distinct physical features. There is wide variability within individuals who have this condition.

- PKU is an inherited error of metabolism caused by a deficiency in the enzyme phenylalanine hydroxylase (PAH) and is now officially referred to as PAH deficiency
- PAH deficiency can result in intellectual disability, organ damage, and neurologic problems
- PAH deficiency is screened for through newborn screening in all 50 states and most developed countries
- Children who have PKU need a Low Protein/Low Phenylalanine (PHE) Diet.
  - PHE is found in all foods that contain protein.
  - Avoid aspartame, an artificial sweetener (i.e. NutraSweet or Equal) containing PHE.
- There is evidence to show that even children who have been on diet since birth are at risk for ADHD and executive function difficulties
- Detailed lists of foods *not allowed*, *allowed but controlled*, and *free food* are not available on-line, however most parents have a listing and a pocket-sized version is available for purchase from Emory University (<http://genetics.emory.edu/clinical/index.php?assetID=309>).

### Recommended Routine Surveillance

- Routine growth monitoring
- Routine monitoring of plasma Phe, Tyr, and plasma amino acid concentrations
- Regular assessment of micronutrient needs
- Mental health evaluation
- Developmental evaluation and therapies as needed
- Monitoring of bone density

Copyright, March 2020; New England Genetics Collaborative / Institute on Disability

[www.gemssforschools.org](http://www.gemssforschools.org)

## Emergency Protocols

- Individuals should have an emergency letter for healthy care providers who may not be familiar with PKU. This letter should detail a detailed explanation of dietary restrictions
- According to the National PKU Alliance (<http://npkua.org/>), public schools are required to make modifications in their meal programs under USDA guidelines for Children with PKU. Be aware, or ask a parent, if the child has a medical alert bracelet.

## Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended. Some of these specialist are only needed for untreated, older individuals

- Geneticist /Genetic Counselor:
  - Diagnosis
  - Coordination of care
  - Genetic risk for family
  - Clinical trials
- Metabolic Multispecialty Clinic
  - Clinical follow-up as needed with a metabolic specialist
  - Dietician familiar with PKU
  - Social worker
  - Nurse specialist
- Neurologist – if needed
  - Epilepsy
  - Parkinson's like features
  - Ataxia
- Orthopedist
  - Bone health assessment
- Developmental Pediatrician
  - Speech therapy
  - Physical therapy
  - Occupational therapy
- Behavior and psychiatric evaluation

## Sample Forms

- Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

Copyright, March 2020; New England Genetics Collaborative / Institute on Disability

[www.gemssforschools.org](http://www.gemssforschools.org)

My patient \_\_\_\_\_ has been diagnosed with PAH deficiency, also known as PKU. This condition is characterized by a lack of an enzyme. Treatment involves not only a special formula containing protein without the amino acid phenylalanine, but also a special low protein diet that includes specially modified foods. When left untreated, PKU can result in intellectual disability, seizures, and behavioral problems.

Because of these restrictions, \_\_\_\_\_ needs the following accommodations. He/she must be allowed access to his/her formula and as this is a condition covered American Disabilities Act, it is required that a reasonable accommodation be made for the student to be able to maintain good dietary management.

## Seven Important Aspects of School Life

[“PKU at a Glance”](#) will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions



## Resources

### GeneReviews: PKU

<https://www.ncbi.nlm.nih.gov/books/NBK1504/>

### New England Consortium of Metabolic Programs

<http://newenglandconsortium.org/> and

<http://newenglandconsortium.org/brochures/educators-guide-to-pku.pdf>

<http://newenglandconsortium.org/toolkit/>

### National PKU Alliance

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

### ACMG Practice Guidelines

<https://www.nature.com/articles/gim2013157>

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3918542/>

Copyright, March 2020; New England Genetics Collaborative / Institute on Disability

[www.gemssforschools.org](http://www.gemssforschools.org)