

PKU At a Glance

PKU stands for phenylketonuria, a metabolic disease in which phenylalanine, which are parts of proteins, cannot be broken down. It is an inherited metabolic condition usually diagnosed in the newborn period through screening in the hospital.

About one in every 15,000 babies is born with PKU.



Meet AJ on page 9

People who have PKU do not have an enzyme to breakdown an amino acid called phenylalanine (PHE). PHE is found in food. When PHE builds up in body tissues, it can prevent normal brain development and can result in an intellectual disability. Early detection and treatment of PKU can prevent severe problems.

PKU is treated with a special diet that must be followed closely in order for children to function well in school and life. Rigidly sticking to the PKU diet and formula is important during the critical period of development in early childhood. It remains necessary throughout adulthood. When children do not follow the diet, they usually do not become physically ill. However, they may have behavioral changes. They will not be able to think as well as when they are following the diet.

Children with PKU do not have any distinct physical features.

Note: There is wide variability within individuals who have this condition.

Things to Think About

1. Medical / Dietary Needs

What you need to know

School staff should treat a child with PKU as a normal healthy member of the class. **However, children who have PKU need a Low Protein/Low Phenylalanine (PHE) Diet.** PHE is found in all foods that contain protein. Therefore, children with PKU need to restrict their intake of protein. Also, avoid aspartame, an artificial sweetener (i.e. NutraSweet or Equal) containing PHE.

Children with PKU often have a special drink. It has most of the protein, vitamins, and minerals that other children get from their foods. The amount of drink and food a child with PKU has daily is carefully calculated by the family and nutritionist. Allow the child to bring their snack/foods into the classroom. They may need access to a refrigerator for special formula. They may also need access to a microwave.

It is extremely important not to allow any food that is forbidden. Even a little taste can result in an increase in PHE levels in the blood. Supervision of younger children with PKU may be needed to prevent sharing or “tastes.”

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. It is important to be sensitive to cultural differences in diet.

Some children may be on a medicine in addition to their diet to help control their PHE levels. This medicine will be administered by the parents.

Be aware, or ask a parent, if the child has a medical alert bracelet.

What you can do

Good communication with parents is very important. Let parents know if a child eats any food not allowed. Also, tell them if the child doesn't eat all the food or formula sent from home. Involve parents to ensure that the student has a safe treat on special days. Parents should send in a supply of low PHE snacks that can be stored in the classroom. Work with cafeteria staff to support the special diet and make it easy for the child to be included.

Explaining Dietary Differences to classmates can be helpful. It is a good idea to involve the family and child in the explanation. A few ideas to think about:

- Children understand the idea of a food allergy.
- Discuss general differences within the class. Emphasize that all people are different.
- People eat different foods (food customs, religious reasons, and regional differences, vegetarian, etc.).
- People have different diets (diabetes, etc.) to help their bodies.
- Involve the school nurse.

Detailed lists of foods not allowed, allowed but controlled, and free food are not available on-line, however most parents have a list and a pocket-sized version is available for purchase from How Much Phe (<https://shop.pkunews.org/products/how-much-phe-the-essentials>).

Children with PKU can always have an apple. It is a PHE-free food.

2. Education Supports

It is important to have HIGH EXPECTATIONS for learning for children who have PKU. Children who have PKU should be treated like any other students, with a low-key approach regarding their medical needs.

What you need to know

Children with PKU may need a 504 plan to accommodate their dietary needs.

- A child may need special arrangements for lunch and for drinking formula during the school day.
- Starting a PHE restricted diet early has eliminated large cognitive delays.
 - However, in spite of early and continuous treatment, children and adults with PKU may experience symptoms. These include cognitive, emotional, and behavioral differences.
- Careful evaluation of any behavioral or learning differences is always important as they may not be related to PKU.

In general, children and adolescents with PKU are more likely than their non-PKU peers to have academic differences. They may need an IEP to be successful in school.

- Some studies show children PKU may differ from controls on full scale IQ, processing speed, attention, inhibition, and motor control.
- Abstract reasoning, executive function, and attention may be areas of weakness.
- PKU that is treated early is associated with average intellectual performance.
- IQ Development is variable.
 - Some individuals with early well controlled diet attain an IQ within an average range. Others may perform within the range of mild to moderate intellectual disability.

Some children with PKU may have difficulties in working memory, motor speed, motor control, sustained attention, and executive function.

- Executive function challenges affect planning, thinking flexibly, and understanding abstract ideas. Children may struggle to remember, process, and organize information efficiently. This can lead to problems in mathematics and reading.
- Executive function is based on a group of interrelated cognitive and behavioral skills. They are responsible for goal directed activity including:
 - Attention
 - Short term memory
 - Planning and organization
 - Behavioral inhibition
 - Social interactions - differences in this area may be due to lack of focus.
- Executive function difficulties may be subtle but present in early and well treated individuals with PKU.
- Executive function and activity levels are affected by PHE levels.
- Cognitive profile is affected by processing speed.
- They may have difficulty remembering locations of objects in space (i.e. number lines may be ineffective).
- Math fractions, geometric shapes and formulas are hard.
- Poor executive function can lead to difficulties in calculations requiring more than one step.
- Word problems may be challenging.

Reading comprehension may become an issue as a child grows older.

- This is often due to executive function difficulties and sustaining attention and focus.
- The lack of these skills can interfere with new knowledge and the ability to master new skills.

Global processing problems and mild mental processing speed may be present.

- Slower processing speed leads to difficulty with comprehension, the ability to complete tasks, and school performance.
- Social relationships may be more difficult so monitor need for any extra support in developing friendships.

Fine motor speed may be decreased.

- Even early treated children may have awkward pencil grasps and poor handwriting.

Math difficulties may be present.

- Math scores are consistently lower than overall achievement scores. This is thought to be due to a combination of visual spatial or perceptual deficits and executive functioning deficits.

Visual spatial difficulties may be present.

- In addition to problems in math, these can lead to difficulty with spelling rules.

What you can do

Executive functioning

- Be flexible and patient.
- Help with organization.
- Provide information in brief, organized, and specific manner.
- A step-by-step approach is most effective, paying attention to the student's abilities.
- Involve adolescents in interventions and foster an atmosphere that enhances independence.

Motor issues

- Allow enough time for tasks like copying letters and figures, which take longer.
- Consider having child work with an occupational therapist.

Language / communication

- Consider using verbal explanations which are often more effective than visual demonstrations, diagrams and models.
- Enhance verbal information with written materials.
- Word problems may be challenging.

Visual-spatial issues

- Allow time for tasks that may take longer or be difficult.
 - Copying letters, figures and geometric shapes
 - Math fractions and formulas
- Individuals need help lining up numbers to do calculations.

Accommodations and modifications

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- Present information in concrete manner.
- Repetition will help children remember basic facts.
- Simplify information presented on worksheets.
- Written homework may need to be modified.
- Computers may be helpful.

3. Behavior & Sensory Support

What you need to know

Children with PKU may have behaviors similar to children with ADHD. Some cognitive difficulties may be a function of ADHD, executive function challenges, processing speed, and/or neuroanatomical abnormalities. Individuals with increased PHE levels have increased ADHD, are more clumsy, more talkative and more hypersensitive than peers.

Social difficulties and emotional problems may be present. In treated individuals, psychiatric problems are increased compared to siblings or children with other chronic problems. Adolescents often have less independence and lower self-esteem. They may have increased frustration and be less achievement orientated.

They often try to enhance their independence by not following their diet.

What you can do

- Monitor for signs of ADHD and talk to the parents if signs are observed. Some children will need ADHD medication; determining which medication works best is the same as for all children.
- Monitor need for supportive interventions like OT, counseling, or behavioral supports.
- Support their independence while supporting their need to follow their diet.
- Follow a consistent break schedule.

4. Physical Activity, Trips, Events

What you need to know

- A child may need to bring special foods on a trip or have at functions. Supports and preparation may be helpful in planning a trip.

- If you live in New England (USA) and qualify, Northeast_Passage offers Therapeutic Recreation and Adaptive Sports programming (nepassage.org).

What you can do

- Make sure parents are involved in planning for trips and functions.
- Ensure transportation/storage of special formula is arranged.
- Ensure volunteers are aware of dietary and processing issues.
- Arrange supervision around any new eating situations. Restrict protein.

5. School Absences and Fatigue

What you need to know

- Although it is important to have metabolic control throughout the lifetime to reduce risks associated with PKU, individuals with PKU do not have increased absences due to their condition.
- Individuals with PKU should not need accommodations for fatigability.

What you can do

- Contact parents if any change is noted.

6. Emergency Planning

What you need to know / What you can do

If a child accidentally has a food with phenylalanine (PHE), it is important to contact the parents. It is not a "911" emergency. However, notify the parent at the end of the day, or in a pre-arranged way.

7. Resources

An Educator's Guide to PKU

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

[An Educator's Guide to PKU](#), from the New England Consortium of Metabolic Programs, has been created for teachers to help support children with PKU at school. The guide includes information about the low-Phe diet, new research on the most common learning challenges in PKU and how they become apparent in the classroom, guidance for educators on what they can do to help, and space to add notes about the child with PKU in your life.

New England Consortium of Metabolic Programs

<http://newenglandconsortium.org/>

The New England Consortium of Metabolic Programs brings together healthcare professionals at all levels involved in identifying and treating individuals with metabolic disorders.

The goals of the Consortium are to disseminate information, collaborate in the development of social support programs and educational materials, support organizations for parents and adults with metabolic disorders, provide training for students in medicine and related fields, jointly develop and conduct research projects, and establish uniform treatment protocols for individuals with metabolic disorders.

National PKU Alliance

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

The National PKU Alliance is a collaboration of PKU community members joining together as a national voice and supporting local efforts to raise PKU awareness and driving advocacy and education, while ultimately looking for a cure. This site includes links to help families find clinics, learn about legislation and PKU research, and find resources.

Thanks to Christine Brown, Executive Director, who helped us review the GEMSS website. (Dec. 2011)

The New England Connection for PKU and Allied Disorders (NECPAD)

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

NECPAD is a non-profit organization that benefits and supports individuals with Phenylketonuria (PKU) and other related disorders and their families. They are linked with the National PKU Alliance, but highlight regional activities and resources.

Thanks to Denise Queally, who helped us review material for this website.

National PKU News

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

This national non-profit organization is dedicated to providing up-to-date information to families and professionals dealing with PKU.

PKU.com

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

This online resource for "all things PKU" includes a focus area for teens, adults with PKU, and parents. It offers advice and support, educational information, recipes, and links to resources and discussion forums for individuals with PKU and family members.

GeneReviews

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

Learn more about the genetics of this form of PKU.

Medline Plus

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

Consumer-friendly information about human genetics from the US National Library of Medicine

8. Meet a Child with PKU!

AJ and His Experiences with PKU

AJ and his twin brother are 7 years old and live in a small New England town in the USA. On the 6th day of their lives, their parents heard the news that AJ had classic phenylketonuria (PKU) and that his twin brother did not. Although his parents had never heard of it, they quickly learned so much through specialty clinics in a large city. After 9 days in the Neonatal Intensive Care Unit to get stronger, AJ was discharged. He was breastfed initially and his parents kept good records of all



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of his intake and output. As he gets older, they have become a bit more relaxed but still keep daily records. That is important so that they can keep track of how much 'PHE' he takes in.

AJ is a sports fanatic with an infectious laugh. He loves team sports and is very active in soccer, baseball, and basketball. He has been riding a bike since he was three years old! He goes on play dates and has friends over for visits. His mother sends snacks that he can eat on the play dates, or lets the parents know ahead of time that he can have any fruit or water. She even had a refrigerator magnet made for close family and friends with his picture surrounded by all the foods he can eat. Their family, friends, and classroom mothers want to know what foods he can eat so they can create snacks that work for ALL kids!

AJ had early intervention services before he turned three and he attended a typical neighborhood preschool with his brother. He is now enrolled in his neighborhood school, is a second grade student, and has a 504 Plan. He does not have an IEP. The 504 plan outlines what school lunches he can eat and indicates which specialty foods the school can order, how much he can have, and how to prepare. His parents worked very hard to get the accommodations he needed written into the plan. His testing at a nearby clinic has him on track for all of his development.

AJ gets most of his nutrition synthetically, with careful attention to PHE levels. He needs to have a special milk (formulated) during the day. AJ has shared that he is embarrassed to have a thermos throughout the day at school and his mother does not want to have him singled out. They are working on strategies to make this of an issue. At birthday parties and social events, curious children might ask him why his pizza is different than theirs. Soon AJ will be able to have answers as he will be learning to take charge of his diet. His mother shared an exciting note about calculating his PHE intake, stating, "There's an 'app' for that!"

His mother has taken Leadership training offered to parents in her state. She has served on a statewide council for children who have health conditions. She also willingly corresponds with other parents worldwide through the PKU list serve.

AJ's mother shared some useful tips for parents and teachers.

For parents:

- Put together a list of what foods are safe and share with family, friends, and the school.

- Deliver a box of food to the classroom filled with snacks he can eat. Then, if an event arises, he can have a snack. His teacher can let the parents know which food he ate so they can calculate his intake for the day.
- Communication notebooks that go back and forth to school are helpful, especially in the preschool years.
- Share the print outs for teachers from the GEMSS website or from other Websites like Children’s Hospital in Boston. <http://newenglandconsortium.org/brochures/educators-guide-to-pku.pdf> or <http://newenglandconsortium.org/for-professionals/teachers-resources/educators-guide-to-pku/>
- Help your child learn how to explain his dietary needs AND really understand why it is so important to keep giving his body and his brain the fuel it needs.

For teachers and school personnel:

- In some ways, PKU is similar to a food allergy, except that it is NOT life-threatening if he has a food he shouldn’t accidentally. She likes to have AJ “lumped into the allergy crowd.” However, some people don’t share this belief.
- Ask parents what would be helpful for them. For example, be open to talking about changes that either notices like fatigue or sluggishness.
- “Melt downs” could just be age related or might indicate higher levels of PHE. There is nothing special diet-wise to do in the moment other than work through it.
- He cannot have the school lunch foods (main entrée) which is why the family pushed for the 504 Plan. This plan explains that he is required to have specialty low protein foods (ordered by the school district). The family has chosen 5 different meals AJ can have and his mother emails the school with the dates and food substitutes for the coming month. AJ buys his school lunches at the same price the other students pay.