

## What is PKU?

Your child has come back positive on the newborn screening for Phenylketonuria, or PKU. What does this mean?

Phenylketonuria (PKU) is an inherited genetic metabolic disorder in which the enzyme required to break down phenylalanine (PHE), a part of protein, is either missing or does not work correctly. As a result of this missing or defective enzyme, individuals with PKU, if untreated, will develop high levels of PHE in their blood. This can affect brain development and function, causing a variety of issues for individuals with PKU. However, these complications can be avoided by following a strict, low-PHE diet.

The PKU diet includes specially manufactured, low-protein foods as well as fruits and most vegetables. People with PKU need some protein in their diets in order to grow muscle and sustain body functions; the amount varies depending on the specific genetic defect. Food items that need to be avoided include meat, fish, poultry, dairy products, baked goods, and eggs. Thus, in order to get the nutrients they need, people with PKU need a special formula supplement containing all of the necessary components of protein without containing any PHE. Food intake needs to be closely monitored, and PHE levels need to be tested regularly.

Parents often experience feelings of loss when they discover that something might interfere with their child's 'normal' life experience.

However, parents may find comfort in the fact that there is treatment available for children with PKU, and that, with appropriate care, these children often have normal intelligence and a normal life expectancy.

## What causes PKU?

PKU is a rare, metabolic disorder caused when both parents have a change or mutation in the gene that encodes for the enzyme needed to break down PHE. In other words, both parents are carriers of PKU but do not have the disorder. This is called "recessive inheritance." When the parents' chromosomes mix, both carriers pass on the mutation, which results in a child with PKU. Studies show that approximately 1 of every 10,000 (NIH) babies will be born with the disorder.

Virtually all cases of PKU are detected during standard newborn screenings. However, it is important to note that even if your child tests positive, it doesn't necessarily mean that they have the disorder. Some children will have a false positive newborn screen (false alarm). Other children may have a mild variant of PKU known as hyperphenylalaninemia, or hyper-PHE, and may require less strict or even no treatment. Your baby's doctor will recommend additional lab tests in order to make a proper diagnosis.

## Where can my child get treatment?

In Wyoming, most children with PKU are referred to the Inherited Metabolic Diseases (IMD) clinic at the Children's Hospital in Denver. Managing your child's diet will be a key part of his or her treatment regimen. However, as many

parents discover, diet for the child with PKU can be very expensive, and many insurance plans do not provide formula coverage. The professionals at the IMD clinic will assist your family in obtaining the coverage you need. State programs may also be available for Wyoming parents who are having difficulty obtaining appropriate coverage. The Kid Care CHIP insurance program (1-877-KIDS-NOW) offers health insurance coverage for children and teens through age 18 that are uninsured and meet income and eligibility guidelines. Additionally, families who do not have coverage because of low income status may ask about Title 19 Health Insurance at their local Department of Family Services office.

## How can I help my child?

Here are some helpful hints that every parent of a child with PKU should know:

- A child with PKU must have frequent finger or heel sticks in order to measure their PHE levels. Parents can learn how to administer these tests at home in order to avoid frequent trips to the clinic.
- Parents can obtain a baby scale that weighs in ounces and kilograms for home use. Again, this will lead to more time at home and fewer trips to the clinic.
- The USDA (United States Department of Agriculture) has laws in place to ensure that your child receives the diet he or she needs while attending public school.
- Parents can obtain a copy of these laws by visiting [www.fns.usda.gov/cnd/guidance/special\\_dietary\\_needs.pdf](http://www.fns.usda.gov/cnd/guidance/special_dietary_needs.pdf). Parents can also visit with the district

nutritionist before the school year begins so he or she can make appropriate arrangements.

- Families should not avoid going out because of a child's diet. A cooler can be very handy when taking trips with a child with PKU. Parents may pack and freeze foods ahead of time, or go to restaurants that are PKU friendly.
- Parents should not try to adapt their diets to match those of their child. A child's strict PKU diet is not healthy for people who metabolize protein properly. Consistently maintaining a healthy diet of their own is the best way for parents to model a healthy diet for their child.

### Where can I learn more?

Visit <http://www.pkunews.org> for resources, books, and recipes for PKU. To network with other parents of children with PKU, consider joining the listserv (an electronic mailing list) maintained by Emory University. To sign up, visit <http://www.listserv.emory.edu/cgi-bin/wa?SUBED1=pkunews-support-I&A=1>

\*Some information contributed by Mid- Atlantic Connection for PKU and Allied Disorders (MACPAD) and Denver Children's Hospital.

### For more information about PKU contact:



Information & Referral for Parents of Children with Disabilities

### Parent Information Center

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# Phenylketonuria (PKU)

Disability Brochure #29



## Characteristics and Coping Strategies

### Parent Information Center

1-307-684-2277

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