The Arc. of New Jersey Family Institute

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Phenylketonuria

What is Phenylketonuria?
Landau-Kleffner syndrome (LKS) is a rare form of epilepsy and usually begins in Phenylketonuria (fen-ul-ke-toe-NU-re-uh), also called PKU, is a rare inherited disorder that causes an amino acid called phenylalanine to build up in your body. PKU is caused by a defect in the gene that helps create the enzyme needed to break down phenylalanine.

Without the enzyme necessary to process phenylalanine, a dangerous buildup can develop when a person with PKU eats foods that are high in protein. This can eventually lead to serious health problems.

For the rest of their lives, people with PKU — babies, children and adults — need to follow a diet that limits phenylalanine, which is found mostly in foods that contain protein.

Babies in the United States and many other countries are screened for PKU soon after birth. Recognizing PKU right away can help prevent major health problems.

What are the symptoms of Phenylketonuria?
Newborns with phenylketonuria initially don't have any symptoms. Without treatment, babies usually develop signs of PKU within a few months.

PKU symptoms can be mild or severe and may include:

- Intellectual disability (formerly called mental retardation)
- Delayed development
- Behavioral, emotional and social problems
- Psychiatric disorders
- Neurological problems that may include seizures
- Hyperactivity
- Poor bone strength
- Skin rashes (eczema)
- A musty odor in the child's breath, skin or urine, caused by too much phenylalanine in the body
- Fair skin and blue eyes, because phenylalanine cannot transform into melanin — the pigment responsible for hair and skin tone
- Abnormally small head (microcephaly)

Severity varies

The most severe form of the disorder is called classic PKU. Children and adults with less severe forms of PKU, in which the faulty enzyme retains some function, have a smaller risk of significant brain damage. But most children with these forms of the disorder still require a special diet to prevent intellectual disability and other complications.

How is Phenylketonuria diagnosed?

Newborn blood testing identifies almost all cases of phenylketonuria. All 50 states in the United States require newborns to be screened for PKU. Many other countries also routinely screen infants for PKU.

If you have PKU or a family history of it, your doctor may recommend screening tests before pregnancy or birth. It's possible to identify PKU carriers through a blood test. A PKU test is done a day or two after your baby's birth. The test isn't done before the baby is 24 hours old or before the baby has ingested some protein in the diet to ensure accurate results.

Treatment options for Phenylketonuria

The main treatment for phenylketonuria includes:

- A lifetime diet with very limited intake of protein, because foods with protein contain phenylalanine
- Taking a PKU formula — a special nutritional supplement — for life to make sure you get enough essential protein (without phenylalanine) and nutrients that are crucial for growth and general health

A safe amount of phenylalanine differs for each person with PKU and can vary over time. In general, the idea is to consume only the amount of phenylalanine that's necessary for normal growth and body processes, but no more. Your doctor can determine a safe amount through:

- Regular review of diet records, growth charts and blood levels of phenylalanine
- Frequent blood tests that monitor PKU levels as they change over time, especially during childhood growth spurts and pregnancy
- Other tests that may be done to assess growth, development and health

Your doctor may refer you to a registered dietitian who can help you learn about the PKU diet.

Long-term complications of Phenylketonuria
Untreated phenylketonuria can lead to:

- Irreversible brain damage and marked intellectual disability within the first few months of life
- Neurological problems such as seizures and tremors
- Abnormally small head (microcephaly)
- Behavioral, emotional and social problems in older children and adults
- Significant health and developmental problems

Resources

- **Mayo Foundation for Medical Education and Research -** www.mayoclinic.org/diseases-conditions/phenylketonuria/basics/definition/con-20026275
- **National Organization for Rare Disorders** - https://rarediseases.org/rare-diseases/phenylketonuria
- **The PKU Foundation -** www.pkufoundation.com