

**FACT SHEET**  
Healthcare Provider

**Phenylketonuria (PKU)**

**Description:**

Phenylketonuria is an autosomal recessive disorder resulting from a defective phenylalanine metabolic pathway. The absence or deficiency of enzyme phenylalanine hydroxylase prohibits the conversion of phenylalanine to tyrosine. This causes phenylalanine and its breakdown chemicals to accumulate in the blood and body tissues. In the “classic” form of PKU, the enzyme that breaks down phenylalanine is completely or nearly completely deficient. Hyperphenylalaninemia means an elevation of phenylalanine in the blood. It is sometimes also used to describe a group of other disorders that may be caused by a partial deficiency of the phenylalanine breakdown enzyme or the lack of another enzyme important to the processing of this amino acid.

**Incidence in General Population:**

1:25,000 live births

**Symptoms:**

With early treatment, normal intelligence and development can be expected. Infants with PKU appear normal at birth and for the first few months of life. Patients with undiagnosed PKU have progressive developmental delay in the first year of life, mental retardation ranging from moderate to severe, seizures, abnormal gait, autistic-like behavior, and an unusual “musky” odor to their urine. Some other commonly observed features in untreated children include microcephaly, prominent cheek and upper jawbones with widely spaced teeth, poor development of tooth enamel, and decreased body growth.

**Diagnosis:**

Newborn screening—Tandem mass spectrometry identifies elevations in blood phenylalanine on dried-blood-spot filter paper. A normal range is 0-124  $\mu\text{mol/l}$ . A second dried-blood-spot filter paper card may be requested by the Newborn Screening Laboratory if the initial screening result is above the normal range. Infants with presumptive positive screening (critical) results require prompt follow up. If this occurred, the clinician would be contacted by the Metabolic Treatment Center. When notified of these results, the clinician should immediately check on the clinical status of the baby and facilitate referral to the Metabolic Treatment Center. The Metabolic Treatment Center will provide consultation and assistance with diagnostic testing.

**Monitoring:**

Individuals diagnosed with PKU require life-long medical management and dietary therapy coordinated by nutrition and metabolic specialists. Clinical observation is important for healthcare providers caring for patients with PKU. It is important for primary care provider and the Metabolic Treatment Center to develop an ongoing collaborative relationship in caring for these patients.

**Treatment:**

Some phenylalanine is essential for normal growth and development. The goal of PKU treatment is to provide the essential amino acids and maintain a low blood level of phenylalanine. This requires a diet that restricts the amount of phenylalanine, but provides all of the other essential amino acids. This means that high-protein foods—such as meat, eggs, milk, and cheese—should be avoided. The diet is protein supplemented with phenylalanine-reduced formula. Blood phenylalanine levels are checked frequently and the diet adjusted accordingly to keep the phenylalanine level within the recommended control range for age. Currently this treatment is recommended for life, particularly for individual with “classic” PKU.

**Illness and Immunizations:**

Fever and illness can cause normal body proteins to break down, the liberation of the body's own amino acids, and ultimately, a rise in the blood phenylalanine level.

**Surgical/Surgical Procedures:**

Discuss any plans for surgical and dental procedures with the Metabolic Treatment Center.

**Growth and development:**

- It is crucial to closely monitor all growth parameters on a regular basis.
- In cases with neurological deficits, the child should be referred to an early intervention program and developmental progress should be closely monitored by both the metabolic team and the primary care provider.



The information provided is offered for general information and educational purposes only. It is not offered as and does not constitute medical advice. In no way are any of the materials presented meant to be a substitute for professional medical care or attention by a qualified practitioner, nor should they be construed as such.

November 2005