



Empowering Children. Supporting Families. Building Brighter Futures.

Resource 073 | Metabolic and hormonal conditions

Phenylketonuria

What it means

Phenylketonuria (PKU) is an inherited metabolic condition in which phenylalanine can build up to harmful levels. Untreated high levels can affect brain development. Newborn screening and early individualized treatment help protect development. Ongoing management is needed even when a child feels well. [1]

Causes and risk factors

PKU usually results from disease-causing changes in both copies of the PAH gene, reducing the body's ability to process phenylalanine. Phenylalanine is found in protein-containing foods. Related conditions can also raise its level, so confirmatory assessment is important. Genetic counseling can explain inheritance. [1,2]

General family education. A concern deserves assessment, but this guide does not diagnose a child or prescribe treatment.



Signs and everyday impact

Babies may have no obvious signs when screening detects the condition. Without effective treatment, high phenylalanine can lead to developmental, learning, behavioral, or neurological difficulties. Symptoms are not a reliable way to judge metabolic control. A child can need treatment while appearing healthy. [1,2]

Assessment and diagnosis

An abnormal newborn screen is followed by confirmatory blood testing and metabolic specialist review. Additional testing clarifies the specific disorder and treatment needs. Regular phenylalanine measurements, growth checks, nutrition review, and developmental follow-up guide care. Screening results alone should not prompt a family-designed diet. [1,2]

Treatment and therapy

Management may include an individually prescribed phenylalanine-controlled diet, medical formula or specialized nutrition, and medicines for eligible children. A metabolic dietitian ensures adequate protein equivalents, energy, and micronutrients. Treatment varies with the specific disorder and response. Families need ongoing monitoring and a plan for illness or changing food intake. [1,2,3]

Support at home

Follow the metabolic team's food, formula, and monitoring plan. Ask for practical guidance on childcare meals, food labels, and travel. Support the child in participating in meals and play without shame. Do not remove all protein or replace medical nutrition with unprescribed products.

When to seek help

Contact the metabolic team promptly after an abnormal newborn screen or difficulty maintaining the prescribed intake. Repeated vomiting, poor feeding, dehydration, or unusual lethargy needs urgent medical advice and the child's illness plan.

Questions for your care team

- What specific metabolic diagnosis has been confirmed?
- Which nutrition and medicine options fit my child?
- How often are blood levels and growth checked?



Podcast preparation

Use this outline to develop a later podcast script.

Suggested opening

Welcome to Mariel Early Intervention. Today we discuss phenylketonuria. Phenylketonuria (PKU) is an inherited metabolic condition in which phenylalanine can build up to harmful levels. This is general education; each child needs individualized care.

Opening: Define the topic and its developmental context.

Understanding: Explain causes and signs without inviting self-diagnosis.

Assessment: Describe evaluation and distinguish screening from diagnosis.

Support: Discuss treatment, therapy, and practical family supports.

Closing: Explain urgent signs and one clear next step.

Misconception to address: Myth: PKU can be managed by simply avoiding all protein. Nutrition must be carefully balanced by specialists.

Before recording: update clinical references and local service details, arrange clinical review, and use fully consented or fictional examples.

Terms in plain language

- Phenylalanine: an amino acid found in protein-containing foods.
- PAH: the enzyme and gene commonly involved in PKU.
- Medical formula: prescribed specialized nutrition used in metabolic treatment.

Sources and further reading

Sources accessed 3 October 2026. Numbered references support the clinical overview. Treatment availability and referral routes vary by country.

[1] About Phenylketonuria (PKU)

<https://www.nichd.nih.gov/health/topics/pku/conditioninfo/Pages/default.aspx>

[2] How do health care providers diagnose phenylketonuria (PKU)?

<https://www.nichd.nih.gov/health/topics/pku/conditioninfo/diagnosed>

[3] What are common treatments for phenylketonuria (PKU)?

<https://www.nichd.nih.gov/health/topics/pku/conditioninfo/treatments>