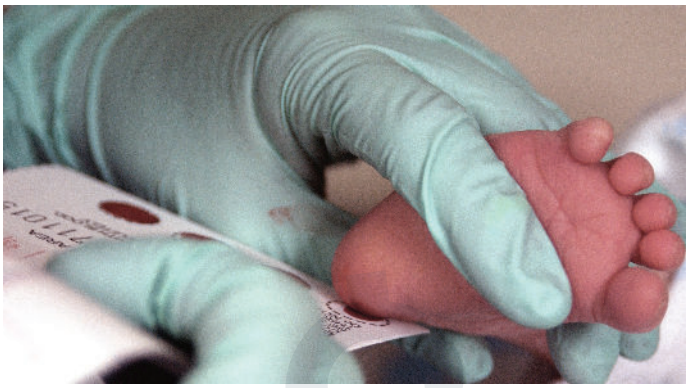
PHENYLKETONURIA PATHOCHART

PATHOPHYSIOLOGY

Phenylketonuria (PKU) is an inherited disorder in which the body cannot metabolize phenylalanine, which is an amino acid found in many foods. When phenylalanine levels get too high, it can cause intellectual disability, brain damage or death. Classic PKU doesn't usually show symptoms until the infant is a few months old, but can lead to severe brain damage if not treated. Early diagnosis and treatment can relieve symptoms and prevent the development of brain damage and intellectual disabilities. Milder forms of the disorder may be controlled with diet.



All newborns undergo testing for Phenylketonuria (PKU), using a drop of blood from the heel.

ASSESSMENT FINDINGS

- Hyperactivity
- Behavioral or emotional problems
- Delayed growth
- Seizures
- Skin rash (eczema)
- Microcephaly

DIAGNOSTICS

- Blood test for enzyme required to break down phenylalanine
  - If not present, positive diagnosis for PKU

NURSING PRIORITIES

- Ensure adequate nutrition
- Patient and family education
- Monitor growth and development

THERAPEUTIC MANAGEMENT

- Monitor growth and development
  - Meat products - red meat, chicken, fish, eggs
  - Dairy products - milk, yogurt, cheese
  - Nuts and legumes
- Seizure precautions

MEDICATION THERAPY

- Special PKU formula for life