NGN NCLEX 2026 · VISUAL STUDY LIBRARY

Phenylketonuria

PKU — Pediatric · Genetics · Metabolic · Nutrition

SYSTEM: Endocrine / Metabolic / Genetics **CLUSTER:** Pediatric Inherited Disorders

NCJMM FOCUS: Recognize Cues · Take Action · Eval Outcomes

📚 **Source Disclosure:** Phenylketonuria (PKU) is not covered in your uploaded project notes. Content below is sourced from standard NGN NCLEX 2026 references (*Saunders Comprehensive Review for the NCLEX-RN, ATI Nursing Care of Children, Lippincott Pediatric Nursing*) and the National PKU Alliance clinical practice guidelines.

01 · DEFINITION

What Is PKU?

An autosomal recessive metabolic disorder where the body cannot break down phenylalanine — leading to toxic CNS accumulation if untreated.

🧬 Genetic Inheritance

Autosomal recessive. Both parents must be carriers. Each pregnancy has a **25% chance** of an affected child.

⚗️ Enzyme Defect

Deficiency of **phenylalanine hydroxylase (PAH)** — the liver enzyme that converts phenylalanine → tyrosine.

📊 Incidence

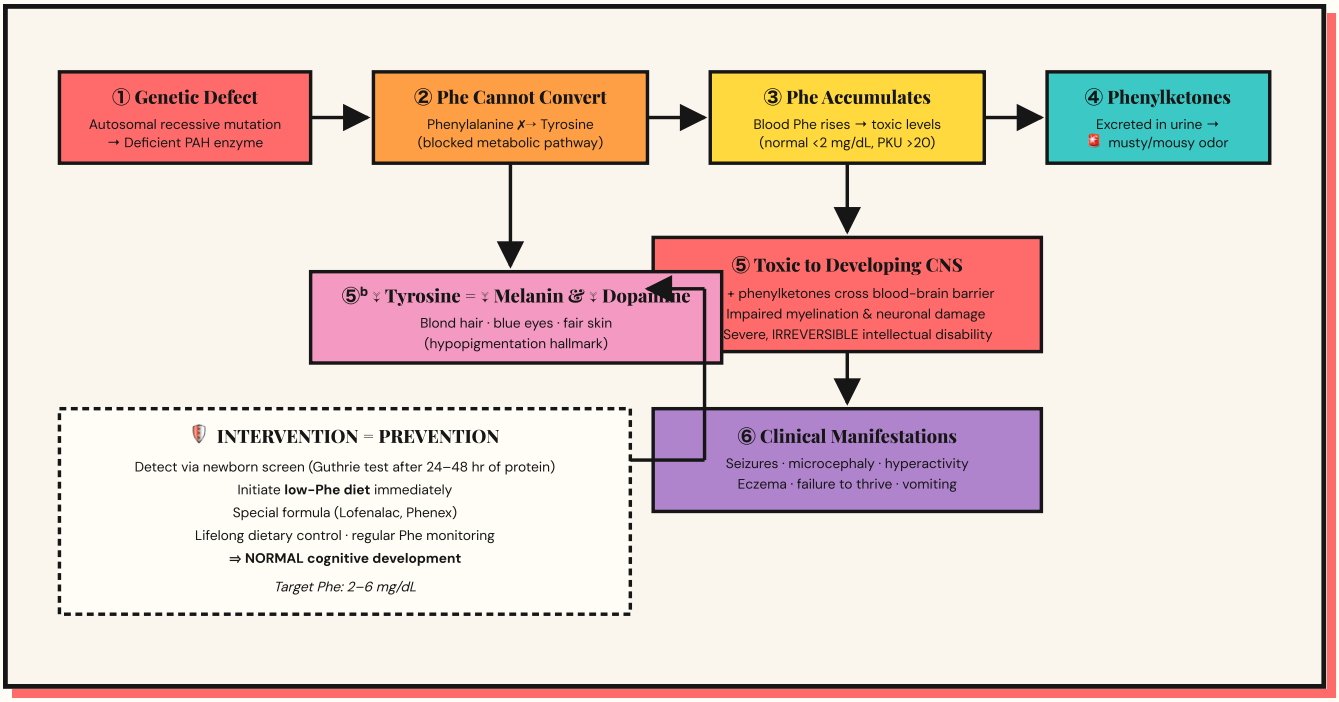
~1 in 10,000–15,000 live births in the U.S. **Mandatory newborn screening in all 50 states.**

WHY IT MATTERS: Untreated PKU causes *severe, irreversible intellectual disability.* Early detection + lifelong dietary management = normal cognitive development. PKU is the #1 example of how newborn screening saves brains.

02 · PATHOPHYSIOLOGY

How PKU Damages the Brain

Trace the cascade: missing enzyme → toxic buildup → neurologic injury → systemic pigmentation changes.



03 · SIGNS & SYMPTOMS

Newborn vs. Untreated Child

PKU is silent at birth. Without screening, signs emerge by 3–6 months as Phe accumulates in the developing brain.

🍼 Newborn / Infant (Early)

- **Asymptomatic at birth** — appears completely normal
- Detected ONLY via newborn screening (Guthrie heel-stick test)
- Must have ingested protein (breastmilk/formula) for **24–48 hours** before testing
- 🚨 **Musty / mousy / barnyard odor** of urine and sweat (phenylketones) — earliest clinical clue
- Mild vomiting and irritability may appear in weeks 2–4
- Eczema or skin rashes (especially diaper area)

🧠 Untreated by 3–6 Months

- 🚨 **Intellectual disability** — severe, progressive, IRREVERSIBLE
- 🚨 **Microcephaly** (small head circumference)
- 🚨 **Seizures** (often by 18 months)
- 🚨 Hyperactivity, irritability, behavioral disturbances
- 🚨 **Hypopigmentation:** blond hair, blue eyes, fair skin (even in families with darker features)
- 🚨 Failure to thrive · vomiting · feeding difficulty
- 🚨 Eczema, dry skin
- 🚨 Awkward gait, jerky movements, hypertonia

🚨 **RED FLAG TRIAD:** Musty urine odor + blond/fair child in a non-blond family + developmental delay = classic PKU presentation if newborn screen was missed.

04 · DIAGNOSIS & SCREENING

The Guthrie Test & Beyond

Timing is everything. Test too early = false negative. Test before protein intake = no Phe to measure.

Newborn Screening (Guthrie Test)

- **Heel-stick capillary blood** on filter paper
- Performed at **24–72 hours of life** (before discharge)
- Infant MUST have ingested **protein** (breastmilk/formula) for at least 24–48 hr first
- If discharged early (<24 hr): repeat screen at 1–2 weeks
- Mandatory in all 50 U.S. states

Confirmation & Monitoring

- **Positive screen** → quantitative serum phenylalanine level
- **Normal Phe:** <2 mg/dL
- **PKU diagnosis:** Phe >20 mg/dL (with normal/low tyrosine)
- **Therapeutic target:** 2–6 mg/dL (lifelong goal)
- Genetic testing confirms PAH mutation
- Lifelong Phe levels drawn regularly to titrate diet

05 · PRIORITY NURSING INTERVENTIONS

ABCs · Safety · Education

PKU isn't an acute crisis — it's a long-game disease where nursing prevents lifelong brain damage through screening + teaching.

🍼 Newborn / Infant

- ✓ **Ensure protein intake** (breastmilk/formula) before Guthrie test
- ✓ Perform heel-stick at 24–72 hr of life
- ✓ If positive screen, refer to metabolic specialist immediately
- ✓ Initiate Phe-free or low-Phe formula (Lofenalac, Phenex-1) right away
- ✓ Breastfeeding may continue but must be **measured + supplemented** with special formula
- ✓ Frequent serum Phe monitoring

👨‍👩‍👧‍👦 Child / Adolescent

- ✓ Maintain **lifelong** low-phenylalanine diet
- ✓ Teach child to self-monitor food choices by school age
- ✓ Monitor growth, weight, developmental milestones
- ✓ Coordinate care with dietitian, metabolic specialist, geneticist
- ✓ Reinforce label-reading skills (aspartame warning!)
- ✓ Address psychosocial impact of restrictive diet (peer pressure, eating out)

⏰ **TIMING IS BRAIN:** Diet must be started within the first *7–10 days of life* to prevent intellectual disability. Every day of delay matters during the brain's rapid growth window.

06 · DIETARY MANAGEMENT

What's On the Plate?

Phenylalanine is an essential amino acid — you can't eliminate it, only restrict it. The diet is high-carb, low-protein, supplemented by special medical formulas.

🚫 AVOID (High Phenylalanine)

- ✕ **Meat, poultry, fish** (all animal protein)
- ✕ **Dairy** — milk, cheese, yogurt, ice cream
- ✕ **Eggs**
- ✕ **Legumes** — beans, lentils, soybeans, peas, peanuts
- ✕ **Nuts & seeds**
- ✕ **Regular bread, pasta, baked goods** (wheat-based)
- ✕ 🧃 **ASPARTAME** (NutraSweet, Equal) — diet sodas, sugar-free gum, sugar-free yogurts, many medications
- ✕ Gelatin, regular protein bars/shakes

✅ ALLOWED (Low Phenylalanine)

- ✓ **Most fruits** (apples, berries, grapes, melons)
- ✓ **Most vegetables** (in measured amounts)
- ✓ **Special low-protein breads & pastas** (medical foods)
- ✓ **Low-Phe medical formulas:** Lofenalac, Phenex-1/Phenex-2, PKU Anamix, Periflex
- ✓ Sugars, jams, jellies, honey, syrup
- ✓ Some fats and oils
- ✓ Special Phe-free baking flours and rice products

⚠ **ASPARTAME WARNING:** Every product containing aspartame in the U.S. is legally required to display *"Phenylketonurics: Contains Phenylalanine."* Teach families to read every label — including chewable vitamins, sugar-free meds, and OTC liquids.

07 · MATERNAL PKU

Special Consideration: Pregnancy

A woman with PKU can damage a non-PKU fetus simply by carrying high blood Phe levels during pregnancy. Pre-conception diet control is non-negotiable.

🤰 The Problem

- Many women with PKU stop their diet as adults (feel "fine")
- High maternal Phe crosses the placenta
- **Fetal Phe is 1.5–2x the maternal level**
- Toxic to developing fetus — even if fetus does NOT have PKU

👶 Fetal Risks

- Microcephaly
- Intellectual disability
- Congenital heart defects
- Intrauterine growth restriction (IUGR)
- Facial dysmorphism

👩‍⚕️ **TEACHING:** Women with PKU must *resume strict diet BEFORE conception* (ideally 3+ months prior) and maintain Phe levels of 2–6 mg/dL throughout pregnancy. This is one of the most testable points on the NCLEX for PKU.

08 · PHARMACOLOGY

Adjunct Therapies

Diet is the cornerstone, but two FDA-approved drugs can help some patients achieve target Phe levels.

DRUG / CLASS	MECHANISM	USE & CONSIDERATIONS	KEY ADVERSE EFFECTS
Sapropterin (Kuvan) Synthetic BH4 cofactor	Provides synthetic tetrahydrobiopterin, the cofactor PAH needs to work — boosts residual enzyme activity	For "BH4-responsive" PKU patients only (≈30%). Oral. Allows liberalization of diet. Approved age ≥1 month.	Headache, rhinorrhea, URI symptoms, diarrhea. Monitor Phe levels.
Pegvaliase (Palynziq) Enzyme substitution	Pegylated phenylalanine ammonia lyase — degrades Phe via alternate pathway	Subcutaneous injection. Adults ≥18 yr only. For uncontrolled Phe levels >600 µmol/L.	🚨 **Anaphylaxis risk** — REMS program required. Inject site reactions, arthralgia.
Medical Formulas Phe-free amino acid mixes	Provide all essential amino acids EXCEPT phenylalanine, plus tyrosine, vitamins, minerals	Examples: Lofenalac, Phenex-1 (infant), Phenex-2 (child/adult), PKU Anamix, Periflex. **Required lifelong.**	Compliance challenges (taste). GI upset if introduced too rapidly.

09 · NCLEX TEST TRAPS

Wrong vs. Right (What Trips Students Up)

High-yield distractor patterns the NCLEX loves to use on PKU questions.

✕ WRONG ANSWER	✓ CORRECT ANSWER
"Perform the Guthrie test immediately after birth before the baby has eaten."	Wait until the infant has had **protein feedings (breastmilk/formula) for 24–48 hours**. Otherwise there's no Phe to detect → false negative.
"The PKU diet can be discontinued once the child reaches adolescence and the brain is fully developed."	The PKU diet is **LIFELONG**. Diet discontinuation in adults causes cognitive decline, mood disturbance, and — critically — fetal damage in pregnancy.
"Diet soda is a healthier choice than regular soda for the child with PKU."	Diet soda contains **aspartame (NutraSweet/Equal)**, which is metabolized to phenylalanine. *Regular soda is actually safer* (within carb limits). This is the most counter-intuitive PKU teaching point.
"Breastfeeding must be stopped immediately when PKU is diagnosed."	Breastmilk has relatively low Phe and can **continue with measurement and supplementation** using Phe-free formula. Total Phe intake is what matters.
"A high-protein diet will help my child grow."	The PKU child needs **low-protein diet** with protein supplied via specialized Phe-free medical formulas. Regular high-protein foods cause irreversible neurologic damage.
"Only the child with PKU needs to follow the diet — the mother with PKU can eat normally during pregnancy."	**Maternal PKU** — mother must resume strict diet *BEFORE conception*. High maternal Phe damages the fetus (microcephaly, heart defects, intellectual disability) even when the fetus doesn't have PKU.

10 · PATIENT & FAMILY EDUCATION

Teaching Points

Empowering families turns PKU from a life sentence into a manageable condition. Education is the #1 nursing intervention.

📖 Daily Living

- **Read every food label.** Look for "Contains Phenylalanine" warning.
- Use a measured/weighed food log; consult metabolic dietitian regularly
- Carry Phe-free formula when traveling, eating out, or at school
- Inform school, daycare, friends, and relatives
- Wear medical alert ID
- Avoid aspartame in ALL forms (gum, mints, sugar-free meds)

🏥 Healthcare Maintenance

- Keep all follow-up appointments with metabolic team
- Regular serum Phe levels (frequency varies by age)
- Genetic counseling for parents (recurrence risk 25%)
- Genetic counseling for child as they reach reproductive age
- Notify all healthcare providers of PKU dx (medications often contain aspartame)
- Pregnancy planning: resume strict diet 3+ months pre-conception

🧠 **NORMAL LIFE IS POSSIBLE:** With early detection and lifelong diet adherence, children with PKU have *completely normal* intelligence, growth, and life expectancy. Reinforce this hope with every family.

11 · MEMORY BOOSTERS

Mnemonics & Pattern Recognition

High-yield associations to lock PKU into long-term memory before exam day.

🐭 "MUSTY MOUSE"

The child smells like a **musty mouse cage** = **PKU**. The mousy/musty urine odor is the single most iconic clinical clue.

👶 "BBB" Triad

Blond hair · **B**lue eyes · **B**rain damage (if untreated). Hypopigmentation from low tyrosine → low melanin.

🥤 "DIET = DANGER"

Counter-intuitively, **DIET sodas are MORE dangerous** than regular sodas in PKU because of aspartame. Read every label.

⏰ "FED before STUCK"

The infant must be **FED protein for 24-48 hr** BEFORE the heel **STUCK** for Guthrie testing — otherwise false negative.

🚫 "MMEEN-Phe" Avoid List

Meat · **M**ilk/dairy · **E**ggs · **E**quae/aspartame · **N**uts/legumes — high in **Phe**.

🤰 "MOM + Phe = FETAL HARM"

Maternal PKU damages a non-PKU fetus. Diet must resume **BEFORE** conception, not after pregnancy is confirmed.

🧬 "25% rule"

Autosomal recessive → each pregnancy of two carriers = **25% affected · 50% carrier · 25% unaffected**.

🎯 "Target 2–6"

Therapeutic Phe goal = **2–6 mg/dL**. Normal is <2, PKU dx is >20. The "two-to-six" range is what dietitians titrate to.

12 · NCJMM CLINICAL JUDGMENT

Six-Step Scenario Walk-Through

Applying the Next-Gen NCLEX Clinical Judgment Measurement Model to a PKU case.

Scenario

A 5-day-old breastfed infant is brought to the clinic for a routine visit. Mother reports the baby is "doing fine." On chart review, the nurse notes the newborn screen drawn at 18 hours of life was reported as *positive for elevated phenylalanine (Phe 28 mg/dL; normal <2)*. Mother is a stay-at-home mom who eats a standard American diet; no family history of PKU is known. The infant is feeding well, has gained weight, and has no obvious symptoms. The faintly musty smell of the diaper area is noted during the exam.

1 Recognize Cues

Elevated Phe on screen · musty diaper odor · early screening timing (18 hr — possibly before adequate protein intake) · no symptoms yet.

2 Analyze Cues

Phe >20 mg/dL with normal tyrosine suggests classic PKU. Musty odor confirms phenylketone excretion. Asymptomatic = early window where intervention prevents damage.

3 Prioritize Hypotheses

(1) Classic PKU — top priority. (2) Maternal/transient hyperphenylalaninemia — rule out via repeat. (3) Possible false-positive due to early draw — confirm with quantitative serum Phe.

4 Generate Solutions

Refer to metabolic specialist STAT · obtain confirmatory serum Phe + tyrosine · initiate Phe-restricted formula (e.g., Lofenalac) · educate parents · continue measured breastmilk feeds · genetic counseling.

5 Take Action

Notify HCP and metabolic team same day · draw confirmatory Phe before initiating any treatment · start Phe-free formula per dietitian within first 7–10 days of life · teach parents how to mix formula and read labels.

6 Evaluate Outcomes

Phe trends toward 2–6 mg/dL · infant achieves normal weight gain · meets developmental milestones · parents demonstrate label-reading and formula prep · genetic counseling completed before next pregnancy.

Sources: Saunders Comprehensive Review · ATI Nursing Care of Children · Lippincott Pediatric Nursing · National PKU Alliance Guidelines