ONCOLOGY · PEDIATRIC · OPHTHALMOLOGY

Retinoblastoma

The most common primary intraocular malignancy of childhood — when a "white glow" in a baby's pupil photograph is a life-and-vision-threatening cancer until proven otherwise.

NGN NCLEX 2026 | NCJMM ALIGNED | HIGH-YIELD | PEDS ONCOLOGY | RB1 GENE · CHROMOSOME 13Q14

📚 **SOURCE DISCLOSURE:** This topic was not present in the uploaded personal notes. Content compiled from standard NCLEX and pediatric oncology references including *Saunders Comprehensive Review for the NCLEX-RN (2026)*, *ATI RN Pediatric Nursing*, *Lippincott Manual of Nursing Practice*, *Hockenberry: Wong's Essentials of Pediatric Nursing*, the *Children's Oncology Group (COG)* retinoblastoma care guidelines, *American Academy of Ophthalmology (AAO)*, and the *American Society of Pediatric Hematology/Oncology (ASPHO)*.

1 Definition & Overview

What It Is

- **Most common primary intraocular cancer of childhood** — arises from immature retinal cells (retinoblasts).
- Driven by mutation/loss of the **RB1 tumor suppressor gene** on chromosome 13q14.
- Median age at diagnosis: **~18 months**; ~95% diagnosed before age 5.

Why It Matters

- **Vision-threatening** AND **life-threatening** — can spread via the optic nerve to the CNS or hematogenously to bone marrow.
- Early detection drastically improves outcomes — survival ≈95% in high-income countries when caught early.
- Hereditary form carries lifelong risk of **secondary cancers** (osteosarcoma, melanoma, soft-tissue sarcoma).

🧬 Hereditary (Germline) Form

`~40%` `BILATERAL COMMON` `YOUNGER ONSET`

- One RB1 mutation inherited; second occurs somatically (Knudson "two-hit").
- Often **bilateral or multifocal**.
- Autosomal dominant; ~90% penetrance.
- **All siblings need screening.**

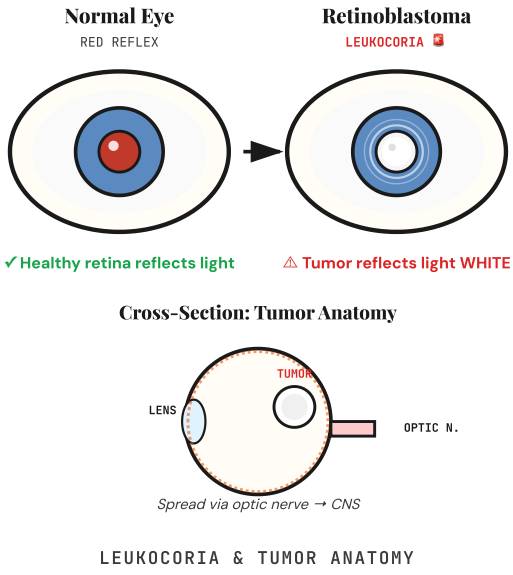
🎯 Sporadic (Somatic) Form

`~60%` `UNILATERAL` `OLDER ONSET`

- Both RB1 mutations occur in the same retinal cell after conception.
- Almost always **unilateral** and **unifocal**.
- Not passed to offspring.
- Typically diagnosed at older age (~24 months).

2 Pathophysiology

1. Normal **RB1 gene** on chromosome 13q14 regulates the G1→S checkpoint of the cell cycle, holding retinal cell division in check.
2. **Knudson "two-hit" hypothesis:** BOTH alleles of RB1 must be inactivated for tumor formation.
3. Loss of RB1 → loss of cell-cycle brake → **unchecked retinoblast proliferation**.
4. Tumor grows **endophytic** (into vitreous) or **exophytic** (under retina → retinal detachment). Mass reflects light → **leukocoria**.
5. Spread routes: **optic nerve → CNS** (worst prognosis), **choroidal invasion → hematogenous** (bone, marrow), or local **orbital extension**.



3 Signs & Symptoms

EARLY / CLASSIC

EARLIEST | RED FLAG

- **Leukocoria** ("white pupillary reflex," "cat's-eye reflex") — the #1 presenting sign. Often first seen in **flash photos**.
- **Strabismus** (esotropia or exotropia) — 2nd most common sign; child **cannot fixate** with the affected eye.
- **Absent red reflex** on exam (white or yellow reflex instead).
- Decreased vision in affected eye (hard to detect in infants).
- Red, irritated eye without obvious cause.

LATE / ADVANCED

LATE | EMERGENT

- **Proptosis** (bulging eye) — extraocular extension.
- **Hyphema** (blood in anterior chamber) or spontaneous hemorrhage.
- **Heterochromia** (different-colored irises).
- Signs of secondary **glaucoma** — pain, photophobia, tearing, enlarged globe (buphthalmos).
- Orbital cellulitis appearance (inflammation, swelling).
- Failure to thrive, weight loss, bone pain (metastatic).
- Neurologic signs if CNS spread (HA, vomiting, ↓LOC).

THE PHOTOGRAPH PEARL

A "glowing" white pupil seen in flash photography is retinoblastoma until proven otherwise. Parents often bring in photographs taken weeks or months earlier showing one white pupil and one normal red pupil. Always take this finding seriously — never reassure that it is a "flash artifact."

4 Priority Nursing Interventions

NCLEX PRIORITY FRAMEWORK: Retinoblastoma is rarely an ABC emergency — instead, prioritize **(1) preserving vision**, **(2) preventing metastasis** via prompt referral and treatment, and **(3) safety** of the unaffected/remaining eye.

🔍 ASSESS

- **Red reflex test** on every well-child visit — absent or white reflex = urgent referral.
- Inspect for **strabismus, leukocoria, asymmetric pupils**; review parent-supplied photographs.
- Assess **vision/tracking**, fixation behavior, and child's response to visual stimuli.
- Family/genetic history — any relatives with childhood eye cancer or RB1 mutation?
- Baseline weight, growth, neurologic status (for CNS involvement).

⚡ ACT

- **Urgent referral** to pediatric ophthalmology (ideally within days, not weeks).
- Prepare child/family for **EUA** (exam under anesthesia), **RetCam imaging**, ocular ultrasound, MRI of orbits/brain.
- Coordinate **genetic counseling and RB1 testing** — affects siblings, future children, and metastatic surveillance.
- NPO per anesthesia protocol before EUA / procedures.
- Administer chemotherapy per protocol; monitor for myelosuppression, infection, and chemo-specific toxicities (see Section 5).

🛡️ PROTECT

- Post-enucleation: monitor surgical site, control pain, prevent infection, support the parents through grief over eye loss.
- **Protect the remaining/unaffected eye** — polycarbonate sports goggles, sun protection, avoid contact sports without protection.
- Hereditary patients: avoid **ionizing radiation** when possible (↑secondary cancer risk).
- Infection precautions during myelosuppression (neutropenic precautions).
- Prevent injury related to monocular vision (depth perception loss).

💜 SUPPORT

- Therapeutic communication — parents experience **shock, guilt** ("we missed it"), and anticipatory grief.
- Age-appropriate explanations for the child; use a doll/teddy to demonstrate enucleation if needed.
- Connect family to **child-life specialist, social work, oncology support groups**, financial counseling.
- Sibling support — they may need testing and feel scared.
- Long-term: school re-entry planning, vision rehab, body-image support for prosthesis.

5 Pharmacology & Treatment Agents

DRUG / CLASS	ROUTE / USE	KEY TOXICITIES	PRIORITY NURSING CONSIDERATIONS
Carboplatin *Alkylating (platinum)*	IV systemic chemo (CEV regimen)	Myelosuppression, **ototoxicity**, nephrotoxicity, N/V	Monitor CBC, renal function, baseline + serial audiology. Aggressive antiemetics.
Etoposide *Topoisomerase II inhibitor*	IV systemic chemo (CEV regimen)	Myelosuppression, hypotension during infusion, **secondary leukemia** risk	Infuse **slowly** (30–60 min) — rapid infusion = hypotension. Monitor BP, CBC.
Vincristine *Vinca alkaloid*	IV systemic chemo (CEV regimen)	**Peripheral neuropathy**, paralytic ileus (constipation), jaw pain, SIADH	**VESICANT** — central line preferred; monitor bowel sounds; stool softeners; assess DTRs/strength.
Melphalan *Alkylating agent*	**Intra-arterial** (ophthalmic artery) or **intravitreal** injection	Local myelosuppression, eyelid edema, retinal/vitreous toxicity	Targeted delivery preserves vision — monitor for systemic absorption; post-procedure eye assessment.
Topotecan *Topoisomerase I inhibitor*	Intravitreal or systemic	Myelosuppression (especially neutropenia), diarrhea	Neutropenic precautions; monitor ANC; assess hydration.
Ondansetron / Granisetron *5-HT_3 antagonist antiemetic*	IV/PO before & during chemo	Headache, constipation, QT prolongation	Schedule **before** chemo + ATC during cycle; monitor ECG with risk factors.

Non-Pharmacologic Treatment Modalities

- **Focal therapy** — cryotherapy (small peripheral tumors) or laser photocoagulation/thermotherapy.
- **Plaque brachytherapy** — radioactive plaque sewn temporarily onto the sclera over the tumor.
- **External beam radiation** — used cautiously, especially avoided in hereditary form due to ↑risk of secondary malignancies in the radiation field.
- **Enucleation** — surgical removal of the eye; reserved for advanced disease (Group D/E), no salvageable vision, or high risk of metastasis. Followed by orbital prosthesis fitting.

6 NCLEX Test Traps

A parent shows you a photograph where one of their 14-month-old's pupils appears white. What is the priority action?

❌ Reassure the parent that this is a common "flash artifact" and recommend retaking the photo.

✅ Perform a red reflex assessment and arrange an **urgent referral** to pediatric ophthalmology — leukocoria is retinoblastoma until proven otherwise.

A 9-month-old has intermittent inward deviation of the right eye. The parents ask if this will "go away."

❌ Reassure that crossing of the eyes is normal until age 1 and reassess at the next well-child visit.

✅ Strabismus persisting **beyond 4–6 months of age** requires evaluation — it is the 2nd most common sign of retinoblastoma and can also cause amblyopia.

A child newly diagnosed with bilateral retinoblastoma is admitted. The parents ask whether their other children should be checked.

❌ "Only your other children with vision problems need to be tested."

✅ Bilateral disease = **hereditary form**. Refer all siblings for ophthalmologic screening and offer genetic counseling/RB1 testing for the family.

A child is receiving carboplatin and vincristine. Which assessment finding is the priority?

❌ A mild headache one day after the infusion.

✅ **Absent bowel sounds** (vincristine → paralytic ileus) OR **new hearing loss** (carboplatin ototoxicity) — both require provider notification.

A child with hereditary retinoblastoma has completed treatment. The parents ask about long-term follow-up.

❌ "Once treatment is complete and surveillance scans are clear, no further follow-up is needed."

✅ **Lifelong surveillance** is required — survivors of hereditary retinoblastoma have markedly increased risk of **secondary cancers** (osteosarcoma, melanoma, soft-tissue sarcoma), especially after radiation exposure.

A toddler is scheduled for enucleation tomorrow. The parents are tearful and ask what to tell the child.

❌ "Don't worry about explaining — they're too young to understand."

✅ Use **simple, concrete, age-appropriate language**, a doll/teddy demonstration, and the child-life specialist. Acknowledge parents' grief; emotional support is part of the priority plan.

7 Patient & Family Education

🏠 At Home

- **Check family photos** — leukocoria often shows up earlier on flash photographs than on direct exam.
- Keep all **ophthalmology appointments**; report any new strabismus, eye redness, vision change, or change in eye appearance.
- **Polycarbonate protective eyewear** for sports and play — protect the remaining/unaffected eye for life.
- Sun protection — UV-blocking sunglasses and wide-brim hats.
- Maintain immunizations on the schedule the oncology team approves (live vaccines may be delayed).

🧬 Genetics & Surveillance

- RB1 genetic testing helps determine if disease is **hereditary or sporadic** — guides sibling and offspring screening.
- Hereditary form: **lifelong cancer surveillance** for osteosarcoma, melanoma, soft-tissue sarcoma — annual physical, skin checks, and prompt evaluation of new bone pain.
- Avoid **unnecessary ionizing radiation** (e.g., elective CT scans) — request MRI when possible.
- Reproductive counseling for survivors planning families.

👁️ Prosthesis Care (Post-Enucleation)

- Conformer placed at surgery; **custom prosthesis** fitted ~6 weeks later as orbit heals.
- Clean prosthesis as taught: remove, rinse with mild soap and water, dry, reinsert — never use alcohol or harsh cleaners.
- Annual polishing and adjustment by ocularist (growing child needs new prosthesis periodically).
- Report drainage, pain, redness, ill-fitting prosthesis to provider.
- Reassure: with prosthesis the eye looks natural; affirm the child's wholeness.

💊 During Chemotherapy

- **Infection precautions** — hand hygiene, avoid sick contacts, fever ≥100.4°F (38°C) = call oncology immediately.
- Report bruising, nosebleeds, pallor, fatigue (myelosuppression).
- Soft toothbrush; avoid live vaccines until cleared.
- Hydration and high-fiber diet to combat vincristine-induced constipation; report no BM ≥48 hours.
- Hearing changes during/after carboplatin — schedule baseline + serial audiology.

8 Memory Boosters & Mnemonics

🐱 "CAT'S EYE"

C · A · T

- **C** Cancer of the retina (childhood)
- **A** Absent red reflex (white pupil = leukocoria)
- **T** Two-hit hypothesis (Knudson, RB1 gene)

👁️ "WHITE GLOW"

W · H · I · T · E

- **W** White pupil (leukocoria) — #1 sign
- **H** Heredity matters (bilateral = familial)
- **I** Inward deviation (strabismus, 2nd sign)
- **T** Tumor of retinoblasts (chromosome 13q)
- **E** Enucleation if advanced; Early referral always

💊 Chemo Side-Effect Pairs

Carboplatin → "Carb" your **ears** (ototoxicity) & marrow (myelosuppression)

Vincristine → "Vin-CONSTI-pation" (paralytic ileus, neuropathy)

Etoposide → Infuse **SLOW** ("E-toposide = Easy does it" — hypotension)

Methotrexate → NOT used here, but recall: avoid aspirin (memory bridge from your drug toxicities deck)

📷 "If it GLOWS, it GOES"

Any white pupil in a flash photograph = **goes to ophthalmology, urgently**.

Counterpart: a healthy retina = **RED reflex** (just like the red-eye in family photos).

One red + one white pupil in a photo is the classic "discovered by Grandma" presentation.

🔢 "RB" = Retino-Blastoma & RB1 gene — same letters, same disease

RB 1 gene on chromosome **13** q14 → think "unlucky 13 hits the eye."

Two-hit hypothesis (Knudson) — both alleles must be lost.

Bilateral = inherited; **Unilateral = usually sporadic**. (But not always — get genetic testing to confirm.)

Pattern recognition: pediatric cancer + eye sign + age <5 → retinoblastoma is at the top of the differential.

9 NCJMM Clinical Judgment Scenario

CASE: A 22-month-old toddler is brought to the pediatric clinic by parents who noticed a "white glow" in the child's right pupil in flash photographs taken over the past 3 months. On exam, the nurse confirms **leukocoria** in the right eye with **esotropia** (inward deviation). The left eye shows a normal red reflex and tracks normally. The child has met all developmental milestones; vital signs are within normal limits for age. The parents are tearful and ask, *"Did we miss something? Will our baby be okay?"*

STEP 1 — RECOGNIZE CUES

What data stands out?

Age <3 years; leukocoria (white pupillary reflex) right eye; absent red reflex on the right; strabismus (esotropia) right eye; symptoms reported as **progressive over months** on photographs; left eye normal; parents emotionally distressed and seeking reassurance.

STEP 2 — ANALYZE CUES

What do these cues mean together?

Leukocoria + strabismus + age <5 = the classic triad of **retinoblastoma** until proven otherwise. Differential includes congenital cataract, Coats disease, persistent fetal vasculature, and toxocariasis — but retinoblastoma is the most dangerous and must be ruled out first. The **unilateral** presentation suggests sporadic disease, but genetic testing is still required to confirm. Progressive change on photos suggests growing tumor mass.

STEP 3 — PRIORITIZE HYPOTHESES

What is the leading concern?

#1 Retinoblastoma — life- and vision-threatening malignancy with risk of optic nerve / CNS / hematogenous spread the longer treatment is delayed. Time-to-treatment directly impacts globe salvage and survival. Secondary priorities: family coping, genetic implications for siblings, ruling out bilateral involvement on detailed exam.

STEP 4 — GENERATE SOLUTIONS

What actions could be taken?

(a) **Urgent same-week referral** to pediatric ophthalmology and oncology; (b) prepare family for **EUA**, ocular ultrasound, **MRI of orbits/brain**; (c) initiate **genetic counseling and RB1 testing**; (d) ensure NPO status when EUA is scheduled; (e) provide emotional support and validate parents' feelings — do NOT reinforce guilt; (f) coordinate child-life and social-work support; (g) arrange screening exam for any siblings.

STEP 5 — TAKE ACTIONS

What does the nurse do now?

The nurse **documents the absent red reflex and strabismus**, notifies the provider, and initiates the **urgent ophthalmology referral**. Parents are taught about the next-step workup, given written instructions and contact numbers, and connected to social work. The nurse uses therapeutic communication — *"You did not miss this. You noticed it, and that's exactly why we're*

acting quickly today." — and offers a referral to the family resource center. NPO instructions are given for any anesthesia-required procedures.

STEP 6 — EVALUATE OUTCOMES

Did the plan work?

Expected outcomes: referral completed within 48–72 hours; diagnosis confirmed and tumor staged (e.g., International Classification A–E); treatment plan initiated (focal therapy ± chemoreduction ± enucleation if Group E); parents demonstrate understanding of the diagnosis and follow-up plan and verbalize decreased guilt; RB1 testing scheduled; siblings screened.
Reassessment: ongoing monitoring of treatment response, vision preservation, family adjustment, and long-term surveillance for secondary malignancies if hereditary.