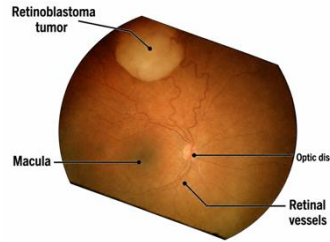


Retinoblastoma is a **primary intraocular malignancy** that rapidly develops from immature retinal cells.

Epidemiology: Retinoblastoma is the most common primary intraocular malignancy of childhood, accounting for 3% of childhood cancers. It usually presents before the age of 5.



Adapted from Aerts et al. (2006). Original image via Wikimedia Commons, CC BY 2.0. Labels added.

PATHOPHYSIOLOGY

Retinoblastoma is caused by mutational inactivation of both alleles of the **retinoblastoma (RB1) tumor suppressor gene**.

PRESENTATION

Common Signs & Symptoms:

▶ **Leukocoria → urgent ophtho referral**

- Leukocoria (white pupillary reflex) ▶
- Strabismus or nystagmus
- Poor visual tracking
- Red, inflamed, or painful eye

Less Common:

- Decreased vision
- Ocular inflammation
- Corneal clouding



Metastatic Disease

- Headache, vomiting, neurologic symptoms
- Orbital or soft tissue mass
- Systemic symptoms (e.g., weight loss)

DIFFERENTIAL DIAGNOSIS

- Coats disease
- Congenital cataracts
- Retinitis
- Persistent fetal vasculature (PFV)

DIAGNOSIS

- Complete physical examination
- Ophthalmologic examination under anesthesia (EUA)
- Ocular ultrasonography
- Optical coherence tomography (OCT) → to detect small tumours or for surveillance)
- MRI of the brain and orbits → to determine tumour size, potential optic nerve involvement, and any associated intracranial tumour

Biopsy contraindicated → risk of tumour seeding ⚠

SCREENING

Prospective parents with a family history of retinoblastoma should be referred for genetic counseling. All children with a family history of retinoblastoma should be screened:

- ✓ soon after birth
- ✓ every 4 months between ages 3-4
- ✓ every 6 months between age 4-5
- ✓ annual screening between ages 5-7



Routine screening is important for all pediatric patients.

- ✓ red reflex testing should be done at every well baby and child visit
- ✓ first eye exam at 6 months of age, again at 2 to 3, and subsequently every year thereafter

MANAGEMENT

- Treatment depends on tumour number/size, proximity to the fovea/optic nerve, and any presence of metastatic disease.
- It can involve focal or systemic treatments.
- Relevant specialties, such as Pediatric Ophthalmology, Oncology, and Genetics, should be involved.

PROGNOSIS & FOLLOW-UP

- Survival >95% in high-income countries
- Increased risk of secondary malignancies in heritable cases
- Lifelong follow-up required

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