🧿 Retinoblastoma Fact Sheet

Cancer Type: Eye Cancer (Intraocular)
Most Affected Group: Infants and young children (typically under 5 years old)

🌐 Website Reference: OcularCancer.com

🔍 What is Retinoblastoma?

Retinoblastoma is a rare eye cancer that begins in the retina, the light-sensitive lining at the back of the eye. It develops when immature retinal cells grow uncontrollably, forming a malignant tumor.

📊 Key Facts:

- Incidence: ~1 in 15,000 to 20,000 live births
- Age of Diagnosis: Most commonly diagnosed before age 2; 90% of cases occur before age 5
- Unilateral vs. Bilateral:
- Unilateral (one eye): ~60% of cases
- Bilateral (both eyes): ~40% (often hereditary)

- Hereditary Cases: ~40% are linked to a mutation in the RB1 gene

🧬 Genetic Information:

- Caused by mutations in the RB1 tumor suppressor gene on chromosome 13
- Hereditary Retinoblastoma:
- Passed from parent to child
- Often involves both eyes (bilateral)
- Higher risk of secondary cancers later in life
- Non-hereditary Retinoblastoma:
- Typically affects one eye (unilateral)
- Caused by a spontaneous mutation

👁️ Signs and Symptoms:

- Leukocoria: A white "glow" or reflection in the pupil (often seen in photos with flash)
- Strabismus: Misaligned eyes (crossed or drifting eyes)
- Eye redness or swelling
- Poor vision or vision loss
- Enlarged pupil or iris color changes
- Eye pain in some advanced cases

🩺 Diagnosis:

- Dilated eye exam by a pediatric ophthalmologist
- Imaging tests: Ultrasound, MRI, or CT scan of the eye
- Genetic testing for RB1 mutations
- Examination under anesthesia (EUA): Often used to assess tumor size and location

⚙️ Staging:

- Intraocular: Tumor confined within the eye
- Extraocular: Tumor has spread beyond the eye (optic nerve, brain, or other organs)
- International Classification (Groups A–E): Helps determine treatment plans and prognosis

🧪 Treatment Options:

- Enucleation: Surgical removal of the eye (used in advanced or non-salvageable cases)
- Chemotherapy: Systemic or localized (e.g., intra-arterial or intravitreal)
- Laser therapy (Photocoagulation)
- Cryotherapy: Freezing the tumor
- Radiation therapy: Plaque brachytherapy or external beam (used less often today)

- Focal therapies: Used to preserve vision in small tumors

🎯 Prognosis:

- Survival Rate (U.S. & developed countries): Over 95% if caught early
- Vision Preservation: Dependent on tumor location and size
- Follow-up Care: Regular monitoring for recurrence or secondary cancers in hereditary cases

❤️ Support & Resources:

- Genetic counseling for families
- Support groups for parents and survivors
- Early screening recommended for siblings or children of survivors
- Prosthetic eye services after enucleation

📌 Key Takeaways:

- Early detection and treatment lead to excellent survival outcomes.
- Leukocoria is often the first visible sign—check flash photos for white reflections.
- Hereditary cases require lifelong monitoring.
- Advances in treatment now focus on saving both life and vision.

For more information and support, visit:

🌐 OcularCancer.com – Your trusted resource for rare eye cancers, survivor stories, and expert-guided care.