

Retinoblastoma

What is retinoblastoma?

Retinoblastoma is the most common primary malignant intraocular tumor in children. Retinoblastoma originates from the retina, which is the light-sensitive internal lining of the eye. One (unilateral) or both (bilateral) eyes may be affected and it typically occurs in children less than 5 years old.

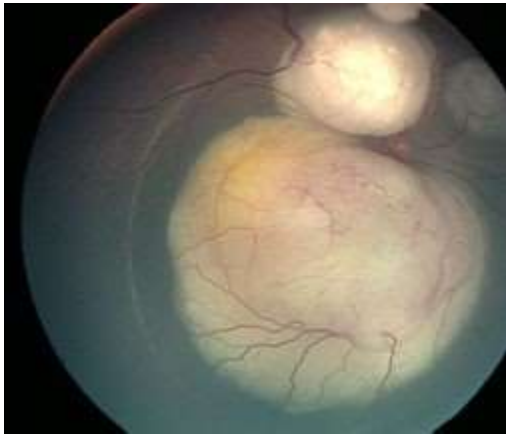


Fig. 1: Retinoblastoma.

How common is retinoblastoma?

Retinoblastoma affects approximately 300-400 children per year in the USA and Canada. Worldwide, about 6000 children develop retinoblastoma each year. It occurs equally in both males and females.

HOW ARE CHILDREN SCREENED FOR RETINOBLASTOMA?

The American Academy of Pediatrics policy statement on Red Reflex Examinations in Neonates, Infants, and Children recommends red reflex testing before discharge from the neonatal nursery and at all subsequent routine health supervision visits. The red reflex refers to the reflection of light off the retina as it passes through the pupil. It is the same red reflex that sometimes makes your pupils red in a standard photograph. An abnormal red reflex requires immediate referral to an ophthalmologist skilled in pediatric examinations, as whitening of the red reflex is the most common presentation for retinoblastoma (referred to as “leukocoria, see Figure 2”). Some

children with retinoblastoma present with [strabismus](#) (lazy eye). All children with strabismus should be evaluated through a dilated eye exam for retinoblastoma. Due to a strong inherited component, screening examinations are recommended for all siblings and offspring of those patients with a history of retinoblastoma (see below).



Fig. 2: An infant with leukocoria of the left eye.

HOW DOES A CHILD GET RETINOBLASTOMA?

The tumor develops when there is a gene abnormality on chromosome number 13. Chromosomes contain the genetic codes that control cell growth and development. The retinoblastoma gene encodes a protein vital to regulating cell growth. 90% of retinoblastoma cases are sporadic and develop “out of the blue” and without warning. 10% have a family member with retinoblastoma. 40% of children with retinoblastoma have a genetic, inherited form of the tumor, even if no one else in the family has the problem. The other type of retinoblastoma is non-inherited and is not passed on from one generation to the next. Retinoblastoma is not caused by external factors such as smoking, drinking, etc.

HOW IS RETINOBLASTOMA DIAGNOSIS CONFIRMED?

An Ophthalmologist examines the eyes either in the office or in the operating room under anesthesia. The ophthalmologist will carefully evaluate the retina after dilating drops are given. Once the patient undergoes complete examination, retinal photography should be performed to document tumor size and assist with staging. Ophthalmic ultrasound should be performed which may show intralésional bright spots consistent with calcium. Due to radiation concerns, CT scans are generally not preferred except in cases that require documentation of calcium and

ultrasonography is not available. Excessive radiation may predispose retinoblastoma survivors to secondary cancers later in life, especially in those patients with a germline mutation. Therefore, MRI is preferred as there is no radiation exposure. Once the child is confirmed to have retinoblastoma, the child should be referred to an oncologist who specializes in the treatment of retinoblastoma and other intraocular tumors.

WHAT IS THE PROGNOSIS FOR RETINOBLASTOMA?

The treatment of retinoblastoma has made enormous strides over the past several decades. Cure rates are upwards of 95% with timely and effective treatment. The prognosis depends on early intervention, the size and spread of the primary tumor, as well as the presence and location of any possible metastatic lesions.

WHAT TREATMENTS ARE AVAILABLE FOR RETINOBLASTOMA?

Treatment of retinoblastoma is tailored to each individual case (Figure 3). The type of treatment depends on a number of factors including laterality, location of the primary tumor, size of the primary tumor, presence of spread beyond the eye (e.g. metastases), and estimated visual prognosis. The management of retinoblastoma is complex and involves strategically chosen methods of surgery (removal of eye when indicated), chemotherapy, and focal therapy. Chemotherapy for retinoblastoma may involve several different methods, including intravenous, intra-arterial, periocular, and intraocular routes. Focal therapy may consist of thermal destruction, freezing (e.g. cryotherapy), laser, and plaque radiation. Single treatment or a combination of treatments may be used. Because of its complexity, retinoblastoma should only be managed by an ocular oncologist trained in the treatment of retinoblastoma. Treatment will usually require a team approach that may involve ophthalmologists, oncologists, geneticists, nurses, social workers, ophthalmologists, and mental health professionals.



Fig. 2: Retinoblastoma post-treatment.

SHOULD SIBLINGS BE INVESTIGATED FOR RETINOBLASTOMA?



All siblings and parents of children with retinoblastoma should have an eye examination by an ophthalmologist. The frequency of screening examinations depends upon age. All newborn babies of affected families need to be screened early in life.

WHAT RESOURCES ARE AVAILABLE TO FAMILIES AFFECTED BY RETINOBLASTOMA?

The impact of the news and the treatment of the disease can affect the entire family. This will be a challenging endeavor for all involved. Support groups are often good resources and there are several useful websites that provide background information.

There are several useful websites that provide background information:

- [The Childhood Eye Cancer Trust](#)
- [The Retinoblastoma Online Support Group](#)
- [Retinoblastoma Survivors Support Group](#)
- [Eye Cancer Heroes](#)

More specific information on support for retinoblastoma patients and their families can be found through your ophthalmologist, AAPOS, and the Children's Eye Foundation of AAPOS. More technical information can be found on the [EyeWiki Site](#).

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