Retinoblastoma

WHAT IS RETINOBLASTOMA?

Retinoblastoma is the most common primary malignant intraocular tumor in children. Retinoblastoma originates from the retina (light-sensitive 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 pre-treatment.

HOW COMMON IS RETINOBLASTOMA?

Retinoblastoma affects approximately 300 children per year in USA and Canada. Worldwide, about 6000 children develop retinoblastoma each year.

HOW IS RETINOBLASTOMA SCREENED?

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. 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.
Some children with retinoblastoma present with strabismus (lazy eye). All children with strabismus should be evaluated through a dilated eye exam for retinoblastoma. Screening dilated fundus examinations on all offspring and siblings of patients with retinoblastoma are recommended.

**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. 90% of retinoblastoma cases 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. Children of affected individuals need exams for retinoblastoma. The other type of retinoblastoma is non-genetic 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 Eye MD examines the eyes either in the office or in the operating room under anesthesia. Children should be screened with fundoscopic examination by an ophthalmologist. Once the patient undergoes complete examination,
fundus photography should be performed to document tumor size and staging. Ophthalmic ultrasound should be performed which may show intrallesional hyperechoic areas (bright spots) consistent with calcium. Computed tomography (CT scan) is generally not preferred for patients with retinoblastoma except in cases wherein a need to document calcification is required and ultrasonography is not available or cannot be done. It is also preferred that CT scan exposure be minimized since children with germline retinoblastoma are at increased risk of developing second cancers and predisposing them to unnecessary radiation may have an additive effect. Therefore, magnetic resonance imaging (MRI) is preferred as there is less radiation exposure. Once the child is confirmed to have retinoblastoma, the child should be referred to an ocular oncologist.

WHAT IS THE PROGNOSIS FOR RETINOBLASTOMA?

With the current modalities of chemotherapy including intravenous, intra-arterial, and intravitreal chemotherapy, cure rates are upwards of 95%. The visual prognosis depends on the size and location of the tumors.

WHAT TREATMENTS ARE AVAILABLE FOR RETINOBLASTOMA?

Management of retinoblastoma should be tailored to each individual. The type of treatment depends on a number of factors including laterality, location of tumor, size of tumor, and estimated vision prognosis.

The management of retinoblastoma is complex and involves strategically chosen methods of enucleation, chemotherapy, and focal therapy. There are 4 routes of delivery of chemotherapy for retinoblastoma including intravenous, intraarterial, periocular, and intravitreal. Focal therapy consists of transpupillary thermotherapy, cryotherapy, argon laser photocoagulation, and plaque radiotherapy. Single treatment or a combination of treatments may be used. Retinoblastoma should be managed by an ocular oncologist trained in the treatment of retinoblastoma.
SHOULD SIBLINGS BE INVESTIGATED FOR RETINOBLASTOMA?

All siblings and parents of children with retinoblastoma should have an eye examination by an Eye MD. The frequency of 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. There is much information from many professionals (Eye MD, Ocularist, Oncologist, etc) and this can be a challenging endeavor. Psychologists, social workers, orthoptists, geneticists and nurses may also aid in the process. Parent contact and support groups are often good resources.

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 technical information can be found on the [EyeWiki Site](#).