Retinoblastoma

Retinoblastoma is a type of cancer found in the retina. It is characterized by loss of heterozygosity on chromosome 13, and is most commonly diagnosed in children. Patients may present with leukocoria, strabismus, or vision loss. Osteosarcoma is an important association to remember with this disease. Diagnosis is made via genetic testing while management strategies include cryotherapy, photocoagulation, chemotherapy, radiation, or surgery.



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Characteristics

Loss of Heterozygosity
Lost Hat-arrow-Z-goat
Loss of heterozygosity (LOH) implies that one allele is missing or mutated such that the remaining allele is the sole contribution for that gene product. It is an important mechanism for several cancers, including retinoblastoma. Two mutational events (M1 and M2) contribute to the inactivation of both alleles of the RB1 gene. M1 can be an initial germline or somatic mutation while M2 may be sporadic.

Chromosome 13
Friday-the-13th-guy standing on Chromosome
Retinoblastoma gene (RB1 or RB) mutation is found in 98% of cases of retinoblastoma. It is a tumor suppressor gene found in the long arm of chromosome 13 at locus 14 (13q14).

Children
Child
Retinoblastoma is the most common primary intraocular malignancy in children. Most cases (95%) are diagnosed in children under five years old. It occurs in 1 out of 15,000 live births in the United States.

Presentation

Leukocoria
Luke-Korea
Leukocoria is described as a whitish reflection of the retina instead of the normal "red reflex". This is due to blocking of the normal retinochoroidal vasculature by the tumor. It is also known as "cat's eye pupil" and can be seen in other diseases like toxocariasis and congenital cataract.

Strabismus
Strawberry-mouse
Strabismus is the other common sign of retinoblastoma. It is characterized by misalignment of the eyes.

Vision Loss
Darkened Eyes
Patients can also present with other less common findings, such as decreased vision, vitreous hemorrhage, ocular inflammation, anisocoria, and glaucoma.

Association

Osteosarcoma
Ostrich-shark-comb
In the hereditary form of retinoblastoma, osteosarcoma is classically seen later in life. Osteosarcoma originates from primitive bone-forming (osteoid producing) mesenchymal cells.

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Diagnosis

Genetic Testing

Jeans and Test-tubes

Molecular/genetic testing is used to identify heritable retinoblastoma in asymptomatic at-risk children. Imaging studies like ultrasound, wide-field photography, CT scan, and MRI can be used to determine tumor, calcification, and adjacent structure involvement.

Management

Cryotherapy

Mr. Cry-O

Cryotherapy works by freezing the tumor, resulting in cell death. It is typically used in two or more freeze-thaw cycles, with a month between cycles.

Photocoagulation

Photo-clogs with Laser

Photocoagulation works by using a laser beam pointed through the pupil. It aims to eliminate tumors by heating them by directing the laser to the blood vessels surrounding and supplying the tumor.

Chemotherapy or Radiation

Chemo-head-wrap and Radiation-radio

Chemotherapy is the pillar treatment of retinoblastoma, which is used to reduce the tumor bulk. It is then combined with other forms of local therapies (cryotherapy, radiotherapy [external beam or plaque], thermotherapy, and photocoagulation). Carboplatin, Vincristine sulfate, and Etoposide Phosphate are the chemotherapy choices used in 3-6 cycles depending on the grade of retinoblastoma.

Surgery

Surgeon

Surgery is indicated in retinoblastoma patients with vision loss, no possibility of storing the eye vision back, and no cure from other treatments. Enucleation is the type of surgery used to remove the entire eye and the optic nerve.

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