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Perspective

Clinical Presentations and Management Strategies of Familial Retinoblastoma

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DESCRIPTION

Retinoblastoma, a malignant tumor of the retina, is primarily known as a sporadic condition occurring in children. However, in a subset of cases, it can manifest within families due to hereditary factors. Familial retinoblastoma, characterized by the inheritance of a mutated *RB1* gene, presents unique challenges in diagnosis, management, and genetic counseling. Understanding the variations in its clinical presentation is significant for early detection and optimal care of affected individuals.

Familial retinoblastoma typically arises from germline mutations in the *RB1* gene, located on chromosome 13q14. The *RB1* gene encodes a tumor suppressor protein that regulates cell cycle progression and inhibits tumor formation. In individuals with a germline *RB1* mutation, there is a predisposition to develop retinoblastoma in both eyes (bilateral disease) or multiple tumors within the same eye (multifocal disease).

The clinical presentation of familial retinoblastoma can vary widely, influenced by factors such as the age of onset, tumor characteristics, and presence of associated genetic abnormalities. While the classic presentation includes leukocoria (white pupil), strabismus (misaligned eyes), and decreased vision, there are several notable variations observed in familial cases.

In familial retinoblastoma, bilateral involvement of both eyes is common, occurring in approximately 80% of cases. The tumors may appear simultaneously or sequentially, with variations in size, location, and degree of intraocular seeding. Bilateral disease poses a higher risk of vision loss and requires coordinated treatment strategies to preserve visual function while controlling tumor growth.

Although familial retinoblastoma often presents with bilateral disease, approximately 20% of cases may manifest as unilateral retinoblastoma in individuals with a family history of the condition. In these instances, the affected eye typically harbors a sporadic somatic mutation in the *RB1* gene, while the contralateral eye remains unaffected or develops sporadic retinoblastoma independently.

Trilateral retinoblastoma represents a rare but significant variant of familial disease characterized by the development of an intracranial Primitive Neuroectodermal Tumor (PNET), typically located in the pineal gland or suprasellar region. Trilateral retinoblastoma most commonly affects children under the age of five and is associated with poor prognosis due to the aggressive nature of the intracranial tumor. While retinoblastoma typically presents in early childhood, familial cases may occasionally manifest later in life, during adolescence or adulthood. Late-onset retinoblastoma is often associated with milder phenotypes and fewer systemic complications compared to pediatric-onset disease. However, delayed diagnosis may result in advanced disease and increased risk of metastasis.

Familial retinoblastoma may exhibit atypical features, including extraocular extension, anterior segment involvement, and calcification within the tumor mass. These variations in tumor behavior and morphology pose diagnostic challenges and necessitate comprehensive ocular examination and imaging studies to accurately assess disease extent and plan appropriate treatment. The diagnosis of familial retinoblastoma relies on a combination of clinical evaluation, imaging studies, and genetic testing. Ophthalmic Examination Under Anesthesia (EUA) remains the standard for assessing tumor size, location, and intraocular dissemination. Imaging modalities such as ultrasonography, Computed Tomography (CT), and Magnetic Resonance Imaging (MRI) aid in detecting extraocular extension and evaluating intracranial involvement in suspected trilateral retinoblastoma cases. Genetic testing plays a central role in confirming the diagnosis of familial retinoblastoma and identifying germline mutations in the *RB1* gene. Molecular genetic techniques, including DNA sequencing and deletion/duplication analysis, enable precise characterization of genetic alterations and facilitate genetic counseling for affected families. Identification of a germline *RB1* mutation carries implications for screening siblings and future generations at risk of inheriting the condition.

The management of familial retinoblastoma requires a multidisciplinary approach involving pediatric oncologists, ophthalmologists, radiation oncologists, and genetic counselors.

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Systemic chemotherapy administered either as primary therapy or as an adjunct to focal treatments such as laser photocoagulation and cryotherapy. Delivery of chemotherapeutic agents directly into the ophthalmic artery to target intraocular tumors while minimizing systemic toxicity.

Used in cases of advanced or refractory disease to achieve local tumor control. Surgical removal of the affected eye may be necessary in cases of extensive intraocular disease or in the presence of neovascular glaucoma refractory to other treatments.