

Retinoblastoma in a Two-Year-Old Child: MRI Features and Diagnostic Value

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Abstract

Retinoblastoma is the most common primary intraocular malignancy of childhood. Early diagnosis and accurate assessment of tumor extension are essential for treatment planning and prognosis. We report the case of a two-year-old child presenting with leukocoria. Orbital MRI demonstrated a well-defined intraocular mass arising from the posterior retina, with low signal intensity on T2-weighted images, predominantly peripheral diffusion restriction, and heterogeneous enhancement after gadolinium administration. An associated retinal detachment appeared as a T2-hyperintense subretinal fluid collection without enhancement. No MRI evidence of optic nerve invasion, scleral breach, or extraocular extension was identified, and no pineal or suprasellar mass was seen. The imaging findings were highly suggestive of unilateral retinoblastoma. This case highlights the value of MRI for evaluating local tumor extent, particularly the optic nerve and extraocular structures, and for screening the intracranial midline structures.

Keywords: Retinoblastoma; Leukocoria; Magnetic Resonance Imaging; Retinal Detachment; Pediatric Intraocular Tumor

Introduction

Retinoblastoma is the most common primary intraocular malignancy of childhood and is usually diagnosed before five years of age [1,2]. The most frequent presenting sign is leukocoria, followed by strabismus; less common manifestations include reduced vision, a red or painful eye, and, in advanced disease, proptosis [2]. Prompt diagnosis is essential because prognosis and management are strongly influenced by local tumor extension, particularly postlaminar optic nerve and extraocular involvement. Fundoscopy and ocular ultrasonography are central to the initial diagnosis, whereas high-resolution contrast-enhanced MRI is the key cross-sectional imaging technique for pretreatment staging. MRI assesses the optic nerve, choroid, sclera, extraocular structures, and intracranial midline regions without exposing the child to ionizing radiation [3,4].

Case Presentation

A two-year-old child was referred for evaluation of leukocoria noticed by the parents. There was no significant personal or family medical history. Ophthalmologic examination demonstrated a right intraocular mass associated with retinal detachment.

Imaging findings

Orbital MRI demonstrated a well-defined intraocular mass arising from the posterior retina. The lesion showed:

- Intermediate signal intensity on T1-weighted images.
- Low signal intensity on T2-weighted images.
- Predominant peripheral diffusion restriction.
- Heterogeneous enhancement after gadolinium administration.

Associated retinal detachment was visualized as a T2-hyperintense subretinal fluid collection without post-contrast enhancement.

No MRI evidence of optic nerve invasion, scleral breach, or extraocular extension was identified. Brain MRI showed no pineal or suprasellar mass.

These imaging findings were highly suggestive of unilateral retinoblastoma.

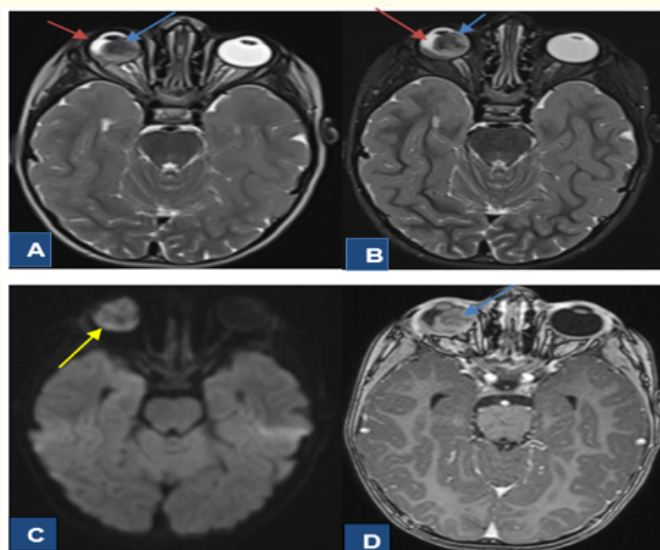


Figure 1: (A, B) Axial T2-weighted and STIR images demonstrate a hypointense intraocular lesion arising from the retina (Blue arrow), with adjacent hyperintense subretinal fluid consistent with retinal detachment (Red arrow). (C) Diffusion-weighted image demonstrates predominantly peripheral diffusion restriction (Yellow arrow). (D) Post-contrast T1-weighted image demonstrates heterogeneous tumor enhancement, without MRI evidence of optic nerve invasion (Blue arrow).

Discussion

Retinoblastoma is primarily driven by biallelic inactivation of the RB1 tumor-suppressor gene in retinal precursor cells and may occur in heritable or nonheritable forms [1]. Unilateral disease is more commonly associated with nonheritable retinoblastoma, although laterality alone does not determine germline status.

Leukocoria is the most frequent presenting sign, followed by strabismus; visual impairment and inflammatory signs may also occur [2]. Early recognition is important for survival and, when feasible, preservation of the eye and vision.

High-resolution contrast-enhanced MRI is the reference cross-sectional imaging technique for pretreatment staging. Its principal roles are assessment of local tumor extension—especially postlaminar optic nerve, choroidal, scleral, and extraocular involvement—and evaluation of the brain for associated midline embryonal tumors [3,4].

Typical MRI findings include an intraocular mass that is relatively hypointense on T2-weighted images, demonstrates diffusion restriction related to high cellularity, and enhances after contrast administration [3,4]. Retinal detachment is a common associated finding and is seen as T2-hyperintense subretinal fluid.

Postlaminar optic nerve invasion, massive choroidal invasion, and extraocular extension are major high-risk features [3,4]. None was identified on MRI in this case; however, MRI cannot exclude microscopic invasion, and histopathology remains the reference standard for microscopic high-risk features when enucleation is performed.

The differential diagnosis of leukocoria includes Coats disease, persistent fetal vasculature, congenital cataract, and medulloepithelioma. MRI contributes to differentiation by characterizing the presence, location, signal, diffusion, and enhancement of a solid intraocular mass and by assessing associated retinal detachment and extraocular structures [5].

MRI in young children also presents important practical and diagnostic limitations. High-resolution orbital imaging is highly sensitive to motion, and sedation or general anesthesia is often required to obtain diagnostic-quality images [4]. MRI is less sensitive than ultrasonography for detecting small intratumoral calcifications; therefore, ocular ultrasonography remains complementary when calcification is diagnostically important [3,4]. In addition, pseudoretinoblastoma conditions—particularly Coats disease and persistent fetal vasculature—may overlap with retinoblastoma on imaging, so MRI findings must be interpreted together with fundoscopy and ultrasonography [5].

Management depends on tumor burden, laterality, visual potential, and the presence of high-risk features, and may include focal therapies, systemic or intra-arterial chemotherapy, intravitreal therapy, and enucleation [1]. MRI is essential for pretreatment staging and may also be used selectively during follow-up according to the clinical context [3,4].

Conclusion

This case illustrates characteristic MRI features of retinoblastoma and emphasizes the role of high-resolution MRI in pretreatment staging. Careful assessment of the optic nerve, choroid, sclera, extraocular tissues, and intracranial midline structures is essential for risk stratification and treatment planning.

Ethics and Consent

Written informed consent for publication of this case and the accompanying images was obtained from the child's parent/legal guardian.

Conflicts of Interest

The authors declare no conflicts of interest.

Authors' Contributions

All authors contributed to the implementation and realization of this work. All authors declare that they have read and approved the final version of this manuscript.

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