

Concomitant Intraocular Retinoblastoma and Choroidal Hemangioma in a 1-Year-Old Boy

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ABSTRACT

We report a case of concomitant choroidal hemangioma in an eye that harbored intraocular retinoblastoma (RB) in a 1-year-old child. A 12-month-old boy presented with right white pupil (Leukocoria). The initial clinical diagnosis was unilateral intraocular RB Group C. The eye was treated initially by systemic chemotherapy combined with focal consolidation therapy followed by I-125 plaque radiotherapy. Massive sub-retinal recurrence was seen 4 months later, and Magnetic Resonance Imaging (MRI) showed extra-scleral invasion of the tumor, and therefore enucleation was performed. Microscopic examination revealed a differentiated retinoblastoma associated at the site of the base of the tumor with choroidal hemangioma with trans-scleral invasion. Orbit MRI was repeated 3 months after the surgery, and there was no orbital tumor recurrence. We are reporting a rare case of pathologically concomitant choroidal hemangioma with trans-scleral invasion in an eye that harbored active recurrent intraocular RB. This trans-scleral extension of hemangioma was misinterpreted by MRI as extraocular retinoblastoma and resulted in enucleation.

Key Words: Choroidal neoplasm, Hemangioma, Leukocoria, Retinoblastoma

INTRODUCTION

Retinoblastoma (RB) is the most common intraocular malignant tumor in children with about two-thirds of cases occurring before the end of the third year of age (1). Such RBs usually remain undetected until they grow large enough to produce a white pupil or result in eye redness and pain due to secondary glaucoma. RB often spreads through the optic nerve, or the orbital tissues by penetrating the sclera (2), and it is therefore not uncommon to see concomitant intraocular and orbital space-occupying lesions in patients with advanced RB. Although the diagnosis of RB is clinical, imaging plays an important role in supporting the diagnosis in addition to the ability to detect extraocular disease extension (4). Magnetic Resonance Imaging (MRI) is used to answer the key clinical questions that may help in the selection of an appropriate line of treatment by detecting tumor extension, optic nerve and retro-bulbar space involvement, as well as brain metastasis (3,4).

Herein we are reporting a very rare case of concomitant choroidal hemangioma with trans-scleral invasion in an eye that harbored active recurrent intraocular RB in a 1-year-old child that was misinterpreted by MRI as extraocular extension of RB. MEDLINE search of the published literature did not show any similar cases.

CASE REPORT

A 12-month-old boy presented with two weeks history of right white pupil (Leukocoria) and loss of fixation when the left eye was closed. There was no family history of eye cancer. On examination, the right eye did not fixate, while the left eye was central, steady and maintained. There was right afferent pupillary defect. No ocular inflammation and no port wine stain were seen.

Examination under anesthesia showed normal anterior segment, and intraocular pressure was 14mmHg in both eyes. Dilated fundus exam of the right eye revealed large macular retinal amelanotic mass associated with focal sub-retinal seeds and vitreous seeds within 3mm of the tumor (Figure 1A). There were no abnormal iris vessels or anterior segment invasion. The left fundus exam was unremarkable.

Orbit MRI revealed a right endophytic, dome-shape intraocular mass that showed a hyper intense signal on T1-weighted images and intermediate signal on T2-weighted images with mild homogeneous post contrast enhancement (0.6cmx1.3cm) and no extra scleral extension or optic nerve invasion.

The initial clinical diagnosis was unilateral intraocular endophytic retinoblastoma (RB); international intraocular retinoblastoma (IIRC) group C (5). The family elected to

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follow conservative therapy (rather than enucleation), and therefore the patient received 6 cycles of systemic chemotherapy (CVE) combined with focal consolidation therapy. Since the tumor was active and localized after 6 cycles, I-125 radioactive plaque was inserted. Initially the tumor showed regression, but tumor recurrence was seen 4 months after plaque surgery, associated with massive recurrent retinal seeds (Figure 1B).

Initial plan for this recurrence was intra-arterial chemotherapy versus enucleation, but new MRI showed extra-scleral invasion at the base of the previously treated tumor (Figure 2A,B). Therefore enucleation was performed. At the time of enucleation, gross inspection of the globe did not show extra-scleral tumor extension or optic nerve invasion. The bisected eye harbored a grossly amelanotic tumor measuring 11x4mm. Microscopic examination revealed a differentiated RB with endophytic growth pattern associated with minimal choroid invasion and no optic nerve invasion or extra scleral extension. The RB cells did not extend into or beyond the sclera, however, there was another amelanotic growth underneath the RB composed of proliferation of thin-walled large caliber vessels (dilated spaces, filled with blood) that had the longest diameter parallel to the surface of the choroid and penetrated the sclera. The growth showed positive staining for CD34 and FLI-1 stains that made it consistent with hemangioma (Figure 3A-D).

Interestingly, this lesion was at the site of the base of the tumor corresponding to the suspicious area of trans-scleral extension seen on MRI, and this had been misinterpreted on MRI as a RB with extraocular extension. There was no histological evidence of extraocular or trans-scleral RB, and the only explanation for the discrepancy between the MRI findings and the pathology findings was the detected unexpected hemangioma at this site.

The final histopathologic diagnosis was differentiated intraocular RB associated with choroidal hemangioma. Orbit MRI was repeated 3 months after the surgery, and there was no tumor recurrence.

DISCUSSION

Orbital extension of RB develops in fewer than 10% of patients and is associated with a higher mortality rate. When the tumor is confined to the globe, the 5 year survival is over 90%, whereas if the tumor extends outside the globe, the mortality is over 90% (6,7). This was the basis for the enucleation decision in this case immediately after the MRI that showed suspicion of extraocular disease extension.

Based upon the findings of a study of 1543 patients with a diagnosis of intraocular and/or orbital space occupying lesions, the differential diagnosis of a lesion involving both the orbit and the eye includes RB, idiopathic inflammatory pseudotumor, lymphoma, and Sturge-Weber syndrome (8).

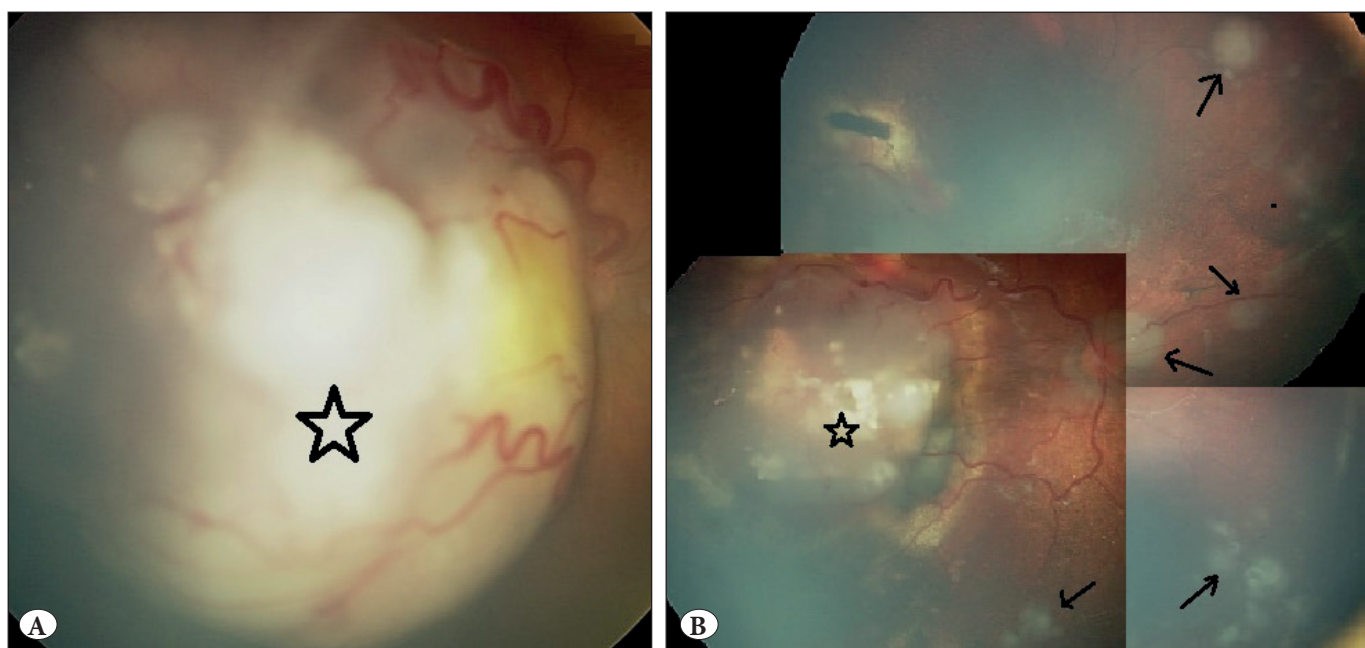


Figure 1: Clinical features. Fundus exam of the right eye at diagnosis **A)** revealed large macular retinal tumor (black star) with enlarged vessels associated with sub-retinal fluid, focal sub-retinal seeds, and vitreous seeds. **B)** Four months after radioactive plaque therapy, the patient developed recurrent tumor activity in the main tumor (black star) associated with multiple active retinal seeds (black arrows).

The most common concomitant scenario is RB with orbital extension, even in a patient with Sturge-Weber syndrome. In a single case in this series, the patient had RB with intraocular and extraocular extension, and although he had Sturge-Weber syndrome, there was no concomitant hemangioma. In our patient, we were looking pathologically for evidence of extraocular tumor extension at the site that was seen in MRI. No extraocular extension by RB was noted in the initial pathologic review of the sections, and therefore we could not find an explanation for the MRI finding of scleral invasive RB. However; upon further pathology review, a hemangioma was noted corresponding exactly to the site and morphology of the scleral invasion reported in MRI. Immuno-histochemical staining confirmed the diagnosis and therefore no further management was necessary. Full body examination of the patient failed to show any evidence of hemangiomas, so this patient did not have Sturge-Weber syndrome.

Choroidal hemangiomas are congenital vascular hamartomas that have been reported in 2 varieties: solitary circumscribed hemangiomas, and diffuse hemangiomas. Riss et al. (9) reported a case of a 4-year-old boy with a diffuse choroidal hemangioma, and without features of the Sturge-Weber syndrome, which presented as a grayish intraocular mass with an overlying retinal detachment associated with orbital mass. The pathologic examination revealed choroidal hemangioma involving the entire choroid with an extra-scleral extension. Similarly, Frau et al. (10) reported a case of atypical circumscribed choroidal hemangioma with retinal detachment in a 41-year-old man in which ultrasonography showed a hyper-echogenic area extending through the sclera, and surgical exploration revealed an extra-scleral hemangioma. Our patient would have the circumscribed type of choroidal hemangioma with trans-scleral extension.

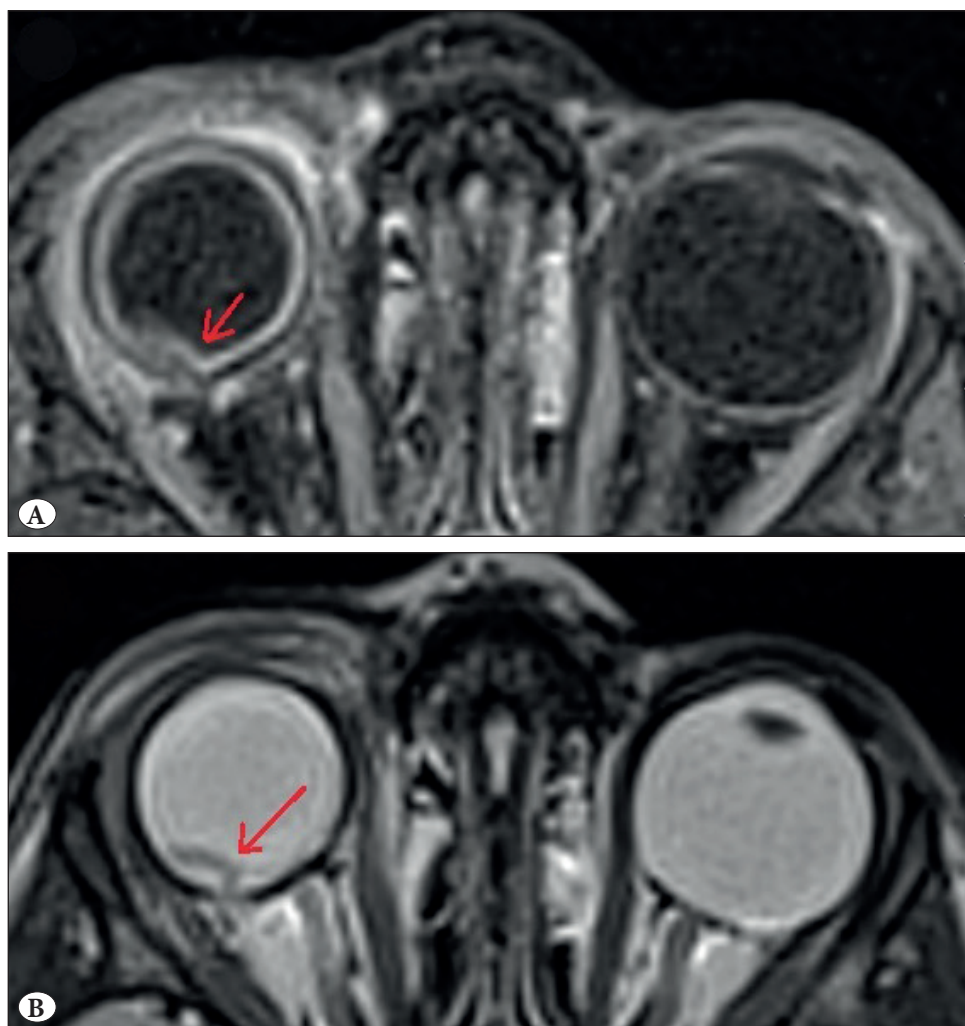


Figure 2: Radiologic findings.

A) Orbit MRI showed a small, contrast enhancing mass in the posterior segment of right eye globe, compatible with clinical features of recurrent tumor (red arrow).

B) There is a focal defect of the overlying sclera associated with thin linear, trans-scleral enhancement extending to extra-ocular tissue (red arrow), which shows intermediate signal on T2WI, similar to the remnant tumor.

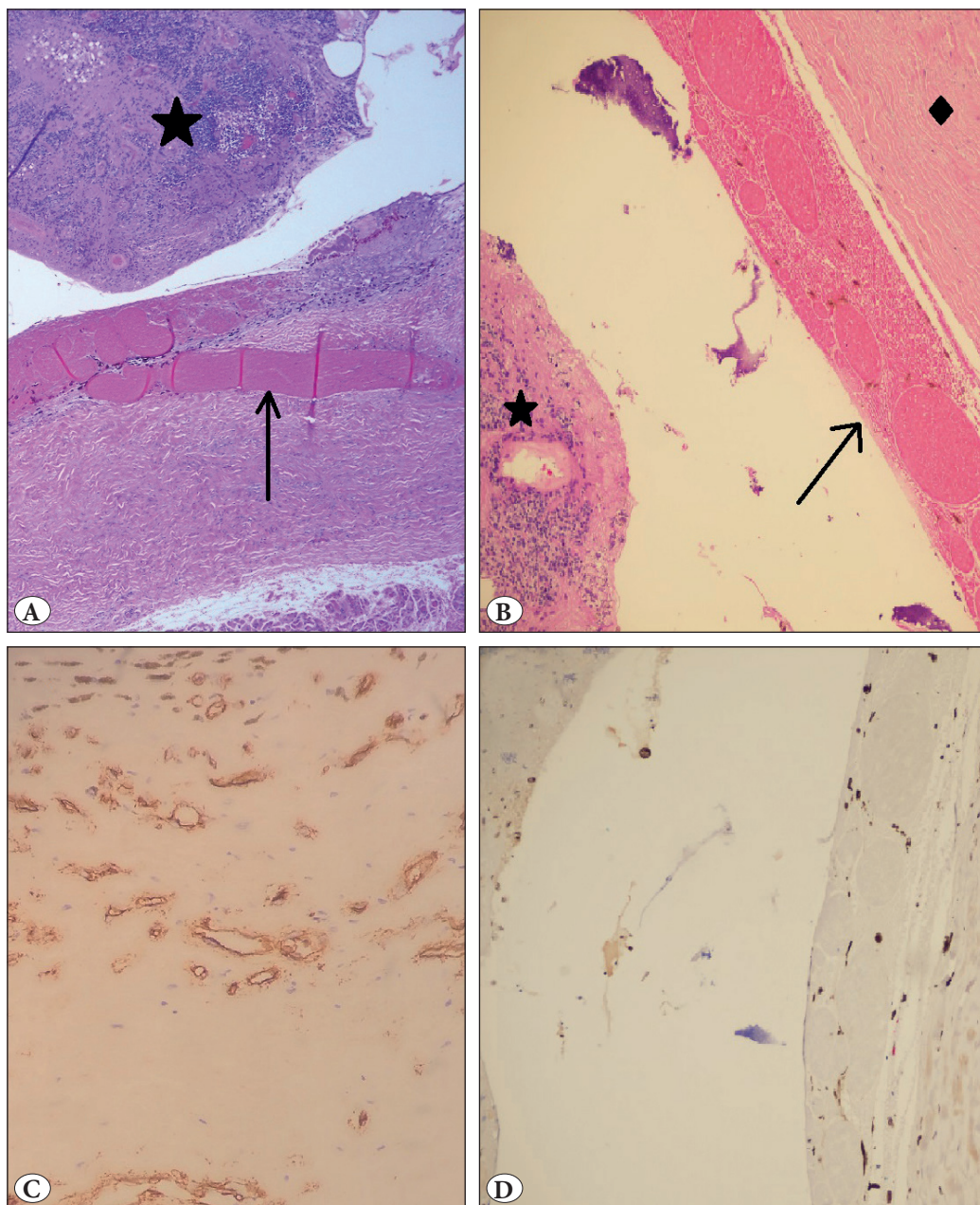


Figure 3: Histopathologic sections showed
A) Retinoblastoma (Black start) overlying another amelanotic choroidal tumor (black arrow) that penetrates the sclera (H&E; x40).
B) This choroidal tumor (black arrow) exists between the Retinoblastoma (Star) and the sclera (Diamond) and consists of dilated spaces, lined by flattened endothelium and filled with blood (H&E; x100). These cells are positive for both **C)** CD34 stain (IHC; x200) and **D)** FLI-1 stain (IHC; x100), which is consistent with hemangioma.

In conclusion we are reporting a very rare case of pathologically diagnosed concomitant choroidal hemangioma with trans-scleral extension in an eye that harbored active recurrent intraocular RB in a 1-year-old child. This trans-scleral extension of hemangioma was misdiagnosed radiologically as extraocular RB and resulted in enucleation. To our knowledge, this is the first case report of this unusual association.

CONFLICT of INTEREST

The authors declare no conflict of interest.

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