

Diffuse Infiltrating Retinoblastoma in a Posttraumatic Contusion Eyeball in a 7-year-old Filipino Male: A Case Report

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ABSTRACT

Diffuse infiltrating retinoblastoma is an extremely rare form of retinoblastoma which is characterized by its atypical growth pattern. This unusual presentation adds complexity to the diagnostic process. The purpose of this paper is to report a rare presentation of diffuse infiltrating retinoblastoma presenting after an ocular trauma.

We described a 7-year-old Filipino boy presenting with total hyphema following an ocular trauma. Comprehensive ophthalmologic clinical and diagnostic evaluations were performed including visual acuity, slitlamp biomicroscopy, ocular ultrasound, neuroimaging, and histopathology post enucleation to determine diagnosis.

The misleading, atypical presentation of diffuse infiltrating retinoblastoma may delay diagnosis. While this dilemma is expected in these scenarios, it should be remembered that timing of diagnosis in retinoblastoma is crucial, as this also equates to optimal management. One should remain vigilant for these uncommon presentations especially in the setting of any intraocular inflammation in children.

Keywords: *diffuse infiltrating retinoblastoma, retinoblastoma, Philippines, case report*

INTRODUCTION

Retinoblastoma is not uncommon in the Philippines.¹ Local data show an incidence of 237/100,000 eye cases.² Recent data also demonstrate the predicted increase in the trend in the number of cases as the years progress.¹ The more common forms of retinoblastoma are the endophytic and exophytic forms. Diffuse infiltrating retinoblastoma is a very rare form of retinoblastoma, and it is characterized by its atypical growth pattern.^{3,4} It presents as a flat, diffusely thickening pattern, without the development of a white vitreous mass typically found in both endophytic and exophytic retinoblastoma. It accounts for less than 1.5-2% of all retinoblastomas.^{3,4} This atypical presentation often provides a misleading presentation and may be confused of more innocuous diseases such as uveitis and other inflammatory disease, which provides more complexity to the diagnostic process and may further delay diagnosis. The most common initial diagnosis is chronic uveitis.^{3,4} Because of its atypical growth pattern, the average age at presentation is 6-7 years and most patients have unilateral involvement.³ In this paper, we report an uncommon presentation of diffuse infiltrating retinoblastoma emerging post traumatically.



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CASE PRESENTATION

A 7-year-old boy was apparently well and had no ocular complaints until six months prior to consult, patient sustained redness of the left eye and absence of vision after being hit by an unrecalled object while he was playing. They immediately sought consult to a local hospital where traumatic hyphema was the recalled diagnosis. They were given medications but offered no relief. It was only six months after the trauma where they consulted our institution. Mother reported no leukocorias or strabismus prior to the injury. Interim history revealed that they consulted different ophthalmologists where a final diagnosis was neither made nor disclosed. There was no family history of any form of cancer in three generations. The patient had no previous hospitalizations, surgeries or co-morbidities. The patient was delivered spontaneously with negative findings on newborn screening. He was fully immunized with routine vaccinations in the local health care facility. No past complaints of loss of appetite and weight.

Patient initially presented with a hyperemic bulbar conjunctiva with an eight ball hyphema with dehemoglobinized blood clots (Figure 1A). Vision of the right eye was 20/40 and no light perception for the left eye. Affected eye was neither ptotic nor proptosed. All extraocular muscles were full and intact in all gazes on duction and version. The pressure of the left eye was 32 mmHg. Slitlamp biomicroscopy revealed a

total hyphema with blood clots without view of the posterior segment. Reverse relative afferent pupillary defect was present. The right eye was unremarkable. Ocular ultrasound revealed diffuse vitreous condensations with low to moderate spikes and no retinal or choroidal detachments were seen (Figure 1B). No calcifications were documented. Our initial impression was a traumatic hyphema, vitreous hemorrhage, and absolute glaucoma from a secondary glaucoma. Patient was started with intraocular pressure-lowering topical medication and was given option for anophthalmic surgery or an anterior chamber washout. They were lost to follow up.

Two months later, patient returned to the clinic and presented with symptoms of body malaise. Mother reported that the eye was gradually proptosing in the interim. Patient presented at the clinic with an 8-mm proptosis of the left eye using Hertel exophthalmometer with marked extraocular movement limitation. There was no view of the pupil due to the diffuse hyphema with suspicious small whitish lesions resembling tumor seeds. Intraocular pressure was elevated to 35 mmHg. Repeat ocular ultrasound showed pockets of lucency and irregular hyperechoicity with high spikes in the vitreous which resemble probable microcalcifications (Figure 1C). The clinical impression of the institution shifted to a masquerade syndrome, probably retinoblastoma, probably a diffuse infiltrating subtype. Immediate admission and MRI of the brain and orbits were performed.

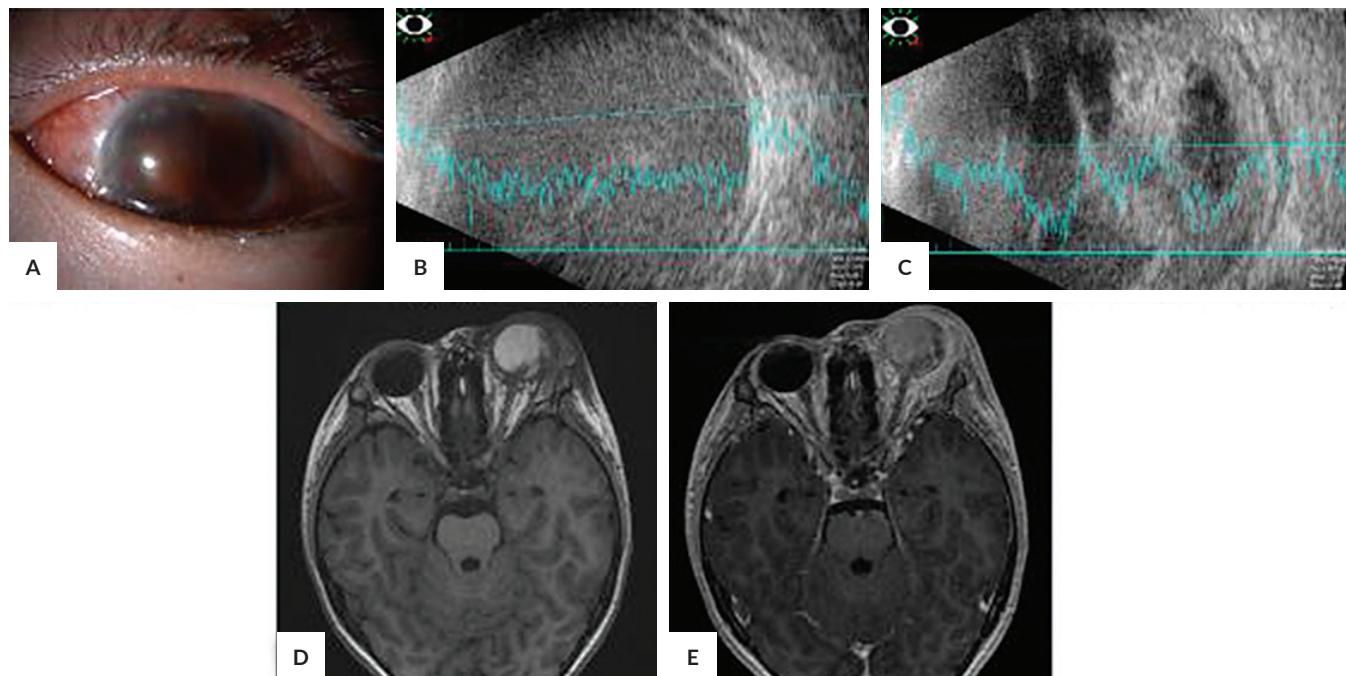


Figure 1. (A) Six months post injury, left eye presented with hyperemic bulbar conjunctiva with an eight ball hyphema with dehemoglobinized blood clots. (B) Ocular ultrasound during the initial consult revealed diffuse vitreous condensations with low to moderate spikes without calcifications or remarkable findings in the retina. (C) Two months after initial consult, ocular ultrasound revealed irregular hyperechoicity with probable microcalcifications Axial T1 sequence with (D) and without contrast (E) revealing flat and irregular thickening of the posterolateral aspect of the retina.

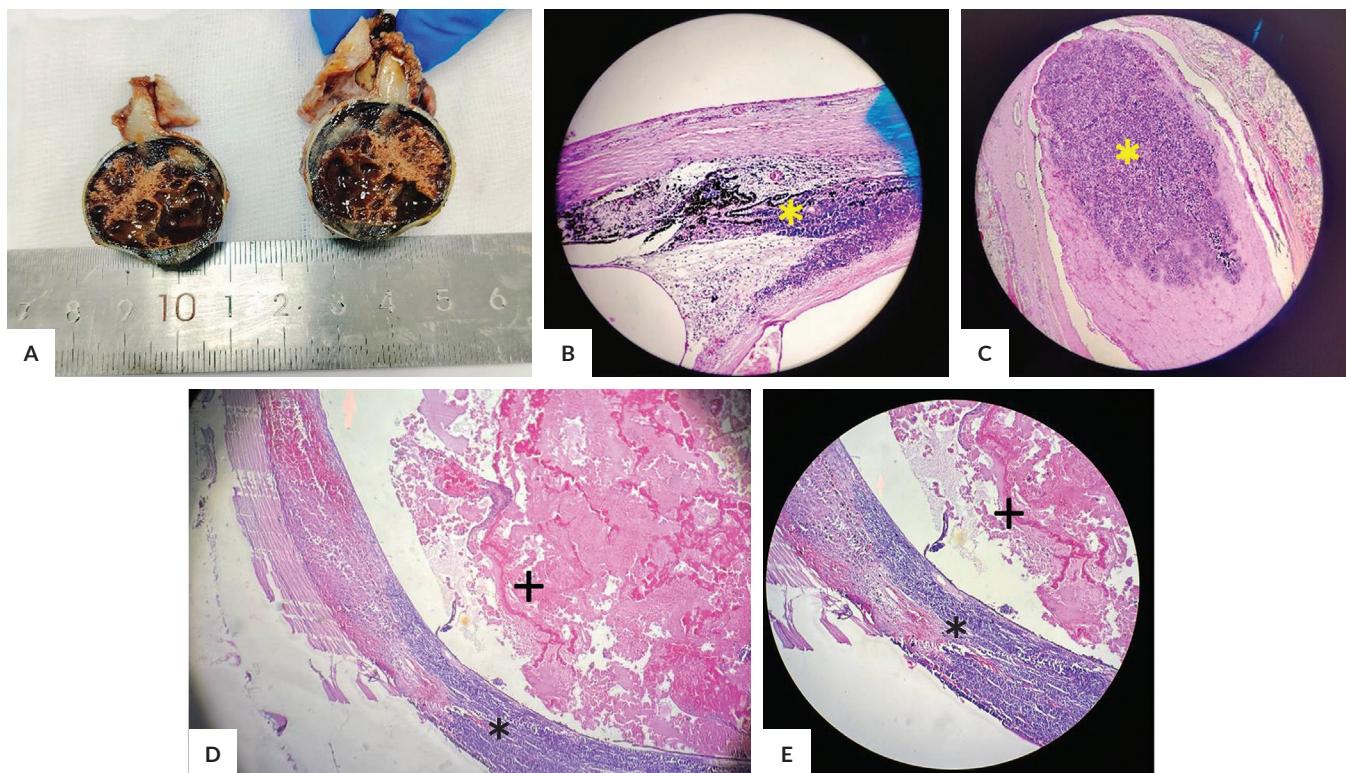


Figure 2. (A) Gross section of the globe showing dark brown cystic surface with tan brown ill-defined necrosis and blood clot formation occupying about 80% of the vitreous cavity. The optic nerve was bulbous and enlarged at the proximal end. Tumor cells (*) were seen infiltrating the iris and ciliary body (B) and involvement of the proximal portion of the optic nerve (C) both at 4x magnification. (D) Tumor cells (*) were seen with diffuse retinal destruction with adjacent blood and necrosis (+) in the vitreous. (E) Tumor cells (*) were seen in the irregular retina arranged in flat nests and tumor necrosis at 40x magnification.

High-resolution orbital and cranial MRI in T1 sequence showed an enlargement of the left eyeball with proptosis and an irregular thickening of the retina in the posterolateral aspect of the eyeball (Figures 1D and E). No vitreous calcifications were identified. Periorbital and retrobulbar soft tissue swelling with diffuse enhancement was demonstrated which was suspicious for extraocular invasion. The left lacrimal gland was also enlarged with enhancement. There were no intracranial masses or enhancements identified.

Parents underwent counseling and the affected eye immediately underwent enucleation and thorough evaluation of the better eye. The fellow eye revealed no lesions. Cut sections of the enucleated eye revealed blood and an irregular brownish-to-fleshy cystic density occupying about 80% of the vitreous cavity, with significant necrosis. An enlarged diameter of the proximal end of the optic nerve was seen (Figure 2A). Microscopically, small hyperchromatic cells with a high nuclear-to-cytoplasmic ratio were seen in the ciliary body (Figure 2B) and spanning the length of the retina (Figures 2D and E) with massive choroidal invasions. The vitreous was occupied with blood and a few monotonous populations of small round blue cells with hyperchromatic nuclei and scant cytoplasm without calcifications. They were

arranged in solid nests of seeds in the vitreous cavity. Other areas showed Homer-Wright rosettes, fleurettes, and few focal calcifications. A post-laminar optic nerve invasion (Figure 2C) was documented. Optic cut nerve end was negative for tumor cells. Histopathologic findings confirm a diagnosis of diffuse infiltrating retinoblastoma.

After enucleation, the parents were counseled for retinoblastoma and the patient was co-managed with pediatric oncology team for systemic surveillance and chemotherapy, and close monitoring and thorough evaluation of the fellow eye was performed.

DISCUSSION

Diffuse infiltrating retinoblastoma is an aggressive pattern of retinoblastoma growth. This pattern demonstrates an eponymous diffuse infiltration of tumor cells along the retina and the absence of an intraretinal or intravitreal mass formation.³ However, tumor formation may occur in diffuse infiltrating subtype and this denotes a later stage of the disease which signifies extensive growth infiltrating the vitreous and other neighboring structures. As opposed to a typical retinoblastoma which presents as a solid mass

formation at the onset in the retina or the vitreous, this form grows diffusely throughout the retina before it may eventually go out to the vitreous. The reason for this unusual growth pattern remains unclear and may mimic other intraocular disease presentation.^{3,4} In many literature, authors have reported delay in the initial diagnosis of this type because of its unusual presentation of a retinoblastoma.¹⁻⁴ The main clinical presentation may be redness, decrease in vision, and pain.² This may be seen clinically as an anterior segment inflammation, pseudohypopyon or an increase in the intraocular pressure.^{2,3} It usually presents with atypical symptoms including decreased visual acuity, pain due to secondary glaucoma, eyelid edema, visible extraocular swelling, or rarely metastasis as the first presentation. Accurate diagnosis presents with difficulty because there are no discrete mass borders and the typical calcification in retinoblastoma is missing. Retinal detachment may often be associated in these cases.

Retinoblastoma is less common in older children, representing only 3.5%–8.5% of cases, and this creates a contrast with diffuse infiltrating type where it is more often diagnosed in older children. The age at presentation is 5.7 years as opposed to typical retinoblastoma which is 12–15 months.³ The growth speed was theorized to be slower in diffuse infiltrating subtype which explains the later age at presentation.³⁻⁵ Ocular signs may vary with the subtype of retinoblastoma. Signs including pseudo-hypopyon, limitation in extraocular muscle motility, hyphema (which may cause a diagnostic dilemma if posttraumatic), iris heterochromia, cataract, secondary glaucoma, and proptosis in later stages of the disease were documented to be atypical presentations of the diffuse type. In the case presented, the clinical picture of the patient fits to the clinical presentation including the age, slitlamp findings, and the delay in the diagnosis that was brought about by the unequivocal clinical and diagnostic findings which posed difficulties in catching the exact diagnosis. Stafford et al. reported that 6.6% of retinoblastoma cases may be misdiagnosed as ocular inflammation.⁶ Higher incidences (17%–34%) of misdiagnosis were found in much older age groups.⁶ A positive family history in diffuse infiltrating subtype is seen in only 4% of cases.^{3,6} Bilaterality is only present in 8% of cases in comparison with typical retinoblastoma which is about 20–30%.⁵

Although diagnostic tools such as ocular ultrasound and MRI are invaluable part of the investigation for retinoblastoma, diagnostic investigation in diffuse infiltrating subtypes do not always confirm the diagnosis.³ Definitive diagnosis may be possible by aqueous humor aspiration.⁴ Recent evidences showed that analysis of tumor cfDNA from aqueous humor may provide a robust method in

cases of inconclusive diagnoses.⁴ Histopathology reveals a diffuse invasion of tumor cells in the retina without tumor formation, or with tumor formation if left untreated.⁷ Lumbar puncture and bone marrow aspiration may be done to determine possible metastasis. Genetic testing plays a role in providing important information for genetic counseling and surveillance.^{4,5} Our case revealed no malignant cells in lumbar puncture and was scheduled for bone marrow aspiration biopsy. Genetic testing is costly in the Philippines, but genetic counseling is performed on the basis of clinical findings.

Due to the highly malignant nature of retinoblastoma, with a mortality rate approaching 99% in the absence of intervention, early diagnosis and treatment are crucial. The primary focus of treatment is patient survival. Secondary goal is the preservation of the globe and its vision. The child was scheduled for follow up at regular intervals while receiving systemic chemotherapy for 6 cycles. While the case presented shows an unsatisfactory delay of eight months to diagnosis, the authors learned a valuable lesson to include retinoblastoma in its differential diagnoses, particularly in the setting of any intraocular inflammation in children with or without history of trauma.

CONCLUSION

The atypical presentation of diffuse infiltrating retinoblastoma often is misleading and may possibly delay diagnosis. While this challenge is expected in such diagnostically-difficult scenarios, it must be remembered that timing of diagnosis in retinoblastoma is crucial, as this also equates to optimal management. One should remain vigilant for these uncommon presentations, and must consider retinoblastoma in their differential diagnoses especially in the setting of any intraocular inflammation in children.

Informed Consent

The study obtained signed informed consent for access to and use of patient data and publication of anonymized patient information.

Statement of Authorship

All authors certified fulfillment of ICMJE authorship criteria.

Author Disclosure

All authors declared no conflicts of interest.

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