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Case Report

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Multiple Misdiagnoses of Retinoblastoma in an 8-Year-Old Boy: A Case Report

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ABSTRACT

Retinoblastoma is the most common intraocular malignancy in children, typically diagnosed before the age of five. Delayed or incorrect diagnosis can lead to extraocular metastasis or intracranial extension. In older children, the presentation may be atypical and mimic conditions such as endophthalmitis, Coats disease, or uveitis, which can make the diagnosis challenging. Here we present a case of unilateral retinoblastoma that was misdiagnosed multiple times, ultimately resulting in intracranial extension and patient death.

An 8-year-old boy with a 7-month history of progressive unilateral proptosis and vision loss was referred for ophthalmologic consultation. He had been misdiagnosed three separate times with uveitis, Coats disease, and optic pathway glioma. Before the correct diagnosis was made, he underwent ventriculoperitoneal (VP) shunt placement for obstructive hydrocephalus. On examination, chemosis, anterior staphyloma, and xanthocoria were noted. The anterior chamber was completely filled with an intraocular mass. Intraocular pressure was 35 mmHg. A previously performed computed tomography (CT) scan showed an intraocular mass with calcification. B-scan ultrasonography revealed a slightly hyperechoic infiltrative mass with fine calcifications. Magnetic resonance imaging (MRI) demonstrated intracranial extension. Cerebrospinal fluid (CSF) cytology was negative for tumor cells. Systemic chemotherapy was initiated immediately, but the patient passed away 3 months later. Successful management of retinoblastoma requires a high index of suspicion to enable early diagnosis. Manifestation after 5 years of age may appear atypical, and mimickers such as endophthalmitis, Coats disease, or uveitis can easily mislead the physician, result in unnecessary interventions, and delay early treatment.

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Introduction

Retinoblastoma is the most common primary intraocular malignancy in children. The average age of diagnosis is 24 months in unilateral cases and 15 months in bilateral cases. Diagnosis after 5 years of age is rare and challenging due to atypical manifestation and sometimes confusing mimickers [1]. A high index of suspicion is essential for early diagnosis and appropriate management before further invasive procedures.

We report a case of unilateral retinoblastoma with intracranial extension that was misdiagnosed three separate times. Additionally, we provide a review of the literature on misdiagnosed retinoblastoma cases published in the PubMed database since 2000, focusing on the initial diagnoses, diagnostic delays, and clinical outcomes.

Case Presentation

An 8-year-old boy presented with a complaint of rapidly progressive unilateral proptosis and was referred to our ophthalmology center for further evaluation. His 5-year-old sister was healthy with no ocular or systemic conditions. Both parents had no notable medical history.

According to the patient's father, the symptoms started 7 months ago with decreased vision and eye deviation. In another country, he was first treated with betamethasone and cycloplegic drops after the diagnosis of uveitis, and then, due to the increase in intraocular pressure (IOP), he was diagnosed as glaucoma secondary to Coats disease and treated with Latanoprost, Brimonidine, and Zilomole drops. Three months later, following the lack of improvement in symptoms and worsening of proptosis, the patient traveled to Iran for further treatment.

During his hospitalization in another center, a VP shunt was implanted for the patient due to significant obstructive hydrocephalus and decreased level of consciousness, to prevent the risk of brain herniation. The initial diagnosis was optic pathway glioma (OPG) at that time.

Upon his first ophthalmic examination, the patient did not respond to the visual acuity exam. However, fixation testing indicated no central, steady, and maintained vision ($C^-S^-M^-$) in the right eye and $C^+S^+M^+$ in the left eye. Relative afferent pupillary defect (RAPD) was positive in the right eye.

In the examination, unilateral proptosis of the right eye along with inward deviation, chemosis, anterior staphyloma, and xanthocoria were evident. An intraocular mass completely filled the anterior segment. The iris and its possible changes were not visible (Figure 1). IOP was measured at 35 mmHg.

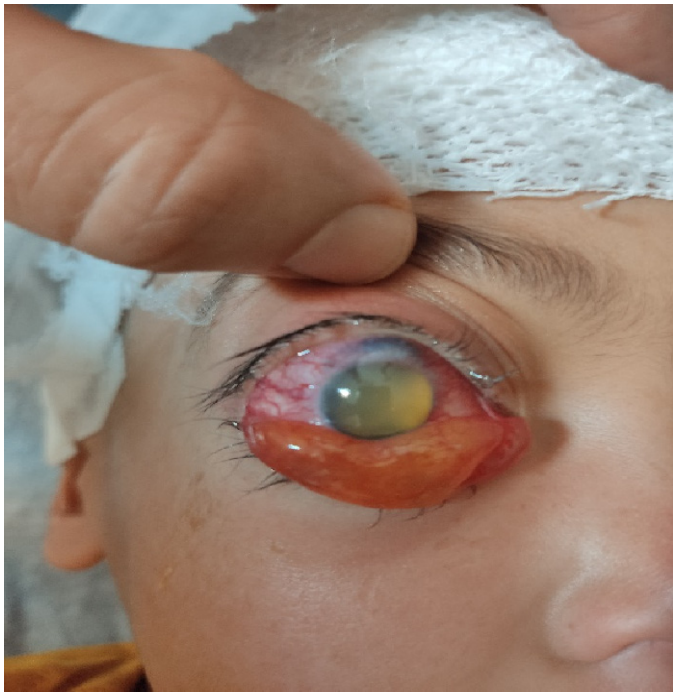

Fig. 1. Picture of patient`s right eye. Severe proptosis with chemosis is noticeable. A moderate sized anterior staphyloma is visible superior to limbus. Intraocular mass completely filled the anterior segment making retinal exam impossible.

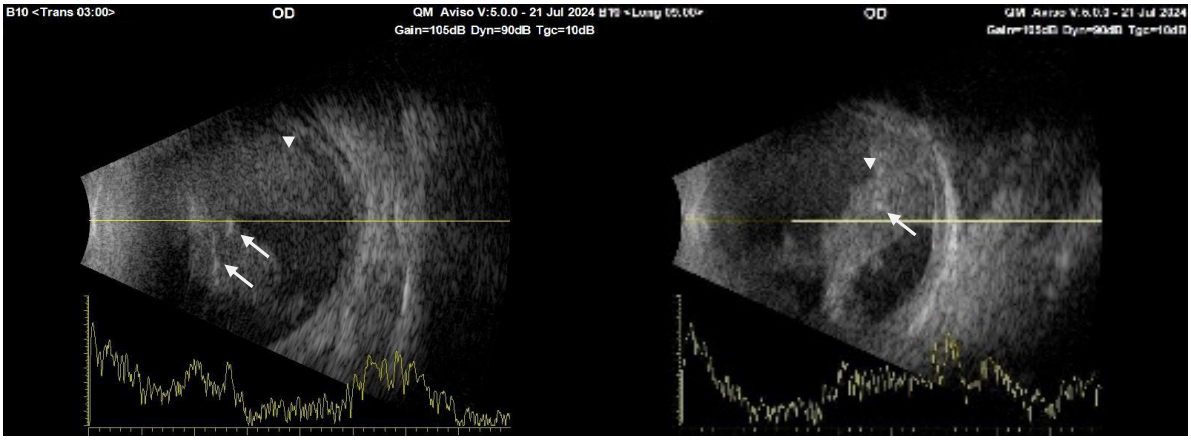

Fig. 2. B Scan ultrasound showing intraocular mass with small hyperreflective points suggesting fine calcifications (white arrows) and adjacent exudative retinal detachment (white arrowhead).

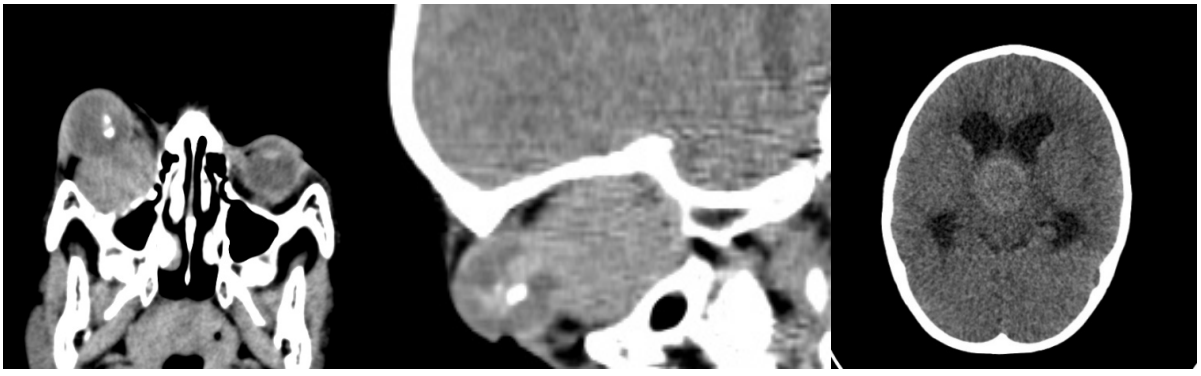

Fig. 3. Orbital and brain CT scan without contrast – soft tissue window (Performed prior to initial ophthalmology consultation): an intraocular infiltrative mass with visible calcification is noted. Also, an intraconal mass extending to the suprasellar region causing obstructive ventricular hydrocephalus is prominent.

Retinal examination was not possible. The left eye anterior segment examination was normal. IOP was measured at 9 mmHg, and optic atrophy was visible. No sign of retinal mass was detected.

B-scan ultrasonography was used to assess intraocular structure. A slightly hyperechoic intraocular infiltrative mass with multiple small internal hyperreflective signals was evident, confirming fine intralesional calcification with exudative retinal detachment (Figure 2). In the previously performed CT scan, an intraocular mass with calcification was seen (Figure 3). In MRI, there was also evidence of choroidal invasion and a retrobulbar mass with a distinct wall extending intracranially through the optic canal, causing obstructive hydrocephalus (Figure 4).

According to the examinations and paraclinical imaging, retinoblastoma with mixed growth pattern along with choroidal invasion and intracranial extension was suggested for the patient. Due to the implantation of a VP shunt, a CSF sample was obtained for tumor cells, which was negative. Systemic chemotherapy was recommended. The patient passed away 3 months after initial treatment.

Discussion

Retinoblastoma is the most common primary intraocular malignancy in children. The prevalence is the same in both sexes. Its incidence is reported from 1 in 15,000 to 1 in 18,000 live births. Forty percent of cases are bilateral [2].

Diagnosis is usually based on fundus examination. The most common symptom is leukocoria. Strabismus, orbital inflammation, retinal detachment, spontaneous hyphema, and pseudohypopyon (tumor cells in the anterior chamber) are other possible symptoms [4].

Suspicious patients should undergo examination under anesthesia (EUA). The location and size of the mass and the presence of vitreous and subretinal seeding should be carefully recorded.

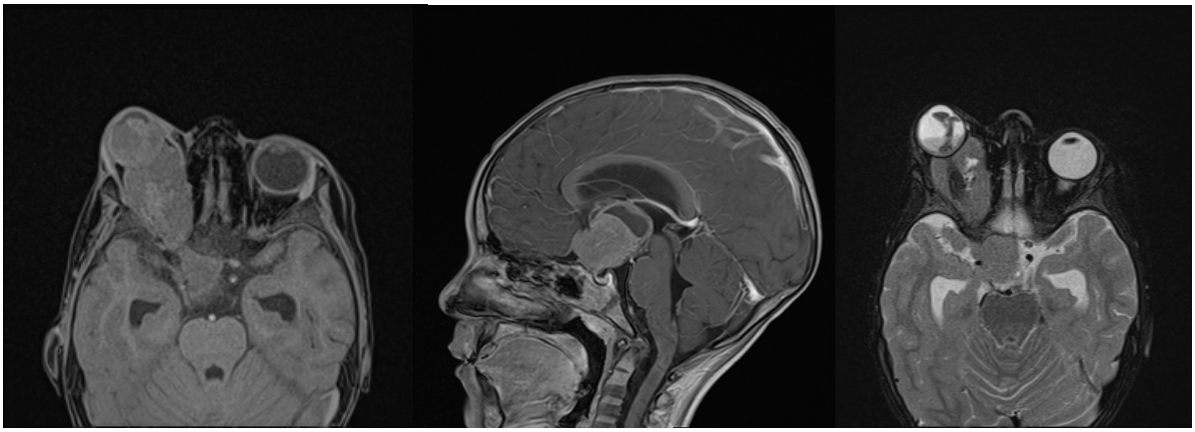

Fig. 4. MRI – with contrast (A) Axial T1 weighted image shows a large intraconal mass with intermediate enhancement and internal hyperintensity, suggesting of necrosis. The vitreous is hyperintense, indicating exudation and areas of hyperintensity again compatible with necrosis. B. Sagittal T1. Intracranial extension via optic canal to supresella. C. T2 weighted image reveals a hypointense intraconal mass relative to the vitreous with heterogeneous signal intensity, consistent with retinoblastoma. Notice choroidal thickening in the right choroid (A, C) suggesting choroidal invasion.

At first, retinoblastoma is usually seen as a round and grayish-white mass, and after it grows, it undergoes necrosis and calcification. Retinoblastoma may grow exophytic, endophytic, or mixed. In exophytic growth, the tumor grows subretinally and leads to exudative retinal detachment and is associated with the risk of choroidal invasion. In endophytic growth, the mass grows into the vitreous cavity and may eventually enter the anterior chamber and cause neovascularization and secondary glaucoma [5].

Retinoblastoma may spread to the central nervous system by invading the optic nerve head. The imaging of choice for investigating CNS involvement is MRI, and it is better to avoid CT scan due to the risk of secondary malignancy following radiation. In patients with extraocular extension and CNS involvement, examination for metastasis with lumbar puncture and bone marrow is recommended. Metastasis may occur through cerebrospinal fluid, choroidal invasion, hematogenous spreading, or conjunctival lymphatics through trabecular meshwork invasion [6].

B-scan ultrasound shows a hyperechoic mass with intralesional calcification seen as hyperreflective spots. However, lack of calcification in older ages can make the diagnosis more challenging. Differential diagnoses include persistent fetal vasculature (PFV), cataract, Coats disease, retinopathy of prematurity, retinal astrocytoma, toxocariasis, medulloepithelioma, etc.

Historically, the treatment of retinoblastoma was limited to enucleation. Over time, globe-salvage treatment techniques including radiotherapy, cryotherapy, intravenous, intra-arterial, and intravitreal chemotherapy were introduced. Also, treatments such as oncolytic viruses and immunotherapy may help control the disease in the future [3].

Here we presented a case of unilateral retinoblastoma in an 8-year-old boy, misdiagnosed multiple separate times as uveitis, Coats disease with secondary glaucoma, and OPG. A CT scan was obtained before asking an ophthalmology consult, which increases the risk of a secondary malignancy. The 7-month delay in diagnosis rendered the tumor inoperable, and due to significant obstructive hydrocephalus, a VP shunt was implanted for the patient, which potentially increases the risk of tumor cell seeding. He also showed optic atrophy in the uninvolved eye, which we assume was the result of prolonged elevated intracranial pressure (ICP). He was referred to an oncologist for further workup and was advised to initiate systemic chemotherapy. He finally passed away 3 months after initial examination. The family did not agree to an autopsy.

A review of 9 reports published in the PubMed database since 2000, comprising 52 patients, revealed that the most common misdiagnoses were endophthalmitis (12 cases), Coats disease [7], traumatic vitreous hemorrhage [6], and uveitis [8].

There is also a report of a 29-year-old man with bilateral multiple retinomas after a spontaneously regressed retinoblastoma. He was followed for 10 years and misdiagnosed as having myopic fundus changes. His daughter was then diagnosed with bilateral retinoblastoma, marking the impact of RB misdiagnosis not only on the patient's early treatment plan but also on early screening of future children [9].

Interestingly, 13 patients had a recent history of ocular trauma prior to diagnosis, raising the question of a possible correlation between ocular trauma and reactivation of spontaneously regressed or arrested retinoblastoma. An ophthalmologist should consider traumatic manifestations as a mimicker of retinoblastoma and look for any sign of ocular mass in ultrasound or other imaging modalities before diagnostic or therapeutic interventions.

Conclusion

Retinoblastoma should be one of the differential diagnoses in cases of posterior uveitis and intraocular malignancy due to the high risk of misdiagnosis or delayed diagnosis and confusing manifestations, especially in older children and even adults.

Ethical Considerations

Study approval statement

Ethical approval was not required for this case report in accordance with local and national guidelines.

Consent to publish statement

Written informed consent was obtained from the patient parents for publication of this case report and accompanying images. A copy of the written consent is available for review.

Authors' contributions

S. K. made the diagnosis and designed the treatment strategy. M. Z. drafted the manuscript, prepared and edited images, gathered clinical data and performed literature review. A. V. and M. N. supervised, reviewed and approved the manuscript. P. K contributed to data collection and manuscript editing.

Data Availability Statement

All relevant data supporting the findings of this case report are included within the article. Additional details are available from the corresponding author upon reasonable request.

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Conflict of Interests

The authors have no conflicts of interest to declare.

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