Masquerade syndrome: Retinoblastoma presenting as phacolytic glaucoma

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1. Introduction

Retinoblastoma is the most common intraocular malignancy of childhood but it is rare occurring in about one in 15000 to 20000 live births[1,2]. Usual presentations of retinoblastoma are leukocoria, strabismus and proptosis[3]. Untreated retinoblastoma is almost always fatal. Therefore, early diagnosis and treatment is critical in saving lives and preserving vision.

2. Case report

A 4-year-old boy presented to hospital with left eye pain for 1 week after being hit by a ball. The pain was intermittent but tolerable by the patient. His mother only noticed left eye redness for the past 1 week and she was unsure any eye redness prior to this episode. She noticed the presence of the whitish mass at the cornea 1 week ago after patient complained of pain. The whitish mass was increasing in size. Patient did not have eye swelling and eye discharge. Prior to this incident, his mother did not notice any presence of leukocoria and squint. Patient did not have any fever, loss of weight and loss of appetite. There was no history of ocular tumour in the family, no recent trauma to the eye or jungle trekking. Patient was born at term with no significant antenatal and intrapartum history. His immunization history is up to date.

On examination, the right vision was 6/6 and left was no light perception. The conjunctiva in his left eye was injected and cornea was edematous. There was pseudohypopyon inferiorly. Lens was opaque and there was no fundus view. Intraocular pressure was 50 mmHg. Patient was initially treated as phacolytic glaucoma with severe inflammation. B scan showed intralesional calcification with retinal detachment. CT orbit showed left intraocular mass with calcification and optic nerve involvement. Thus, our final diagnosis was left eye retinoblastoma. Patient was performed a enucleation with chemotherapy in left eye. One year after the treatment, there was no local and distant invasion of the tumor. Anterior inflammation is a rare form of masquerade syndrome associated with retinoblastoma.

We aim to report an uncommon presentation of retinoblastoma and its management. A 4-year-old boy presented with left eye pain for 1 week with eye redness after being hit by a ball. On examination, the visual acuity in his right eye was 6/6 and left eye was no perception to light. The conjunctiva in his left eye was injected and cornea was edematous. There was pseudohypopyon inferiorly. Lens was opaque and there was no fundus view. Intraocular pressure was 50 mmHg. Patient was initially treated as phacolytic glaucoma with severe inflammation. B scan showed intralesional calcification with retinal detachment. CT orbit showed left intraocular mass with calcification and optic nerve involvement. Thus, our final diagnosis was left eye retinoblastoma. Patient was performed a enucleation with chemotherapy in left eye. One year after the treatment, there was no local and distant invasion of the tumor. Anterior inflammation is a rare form of masquerade syndrome associated with retinoblastoma.

Figure 1. Left eye injected conjunctiva, oedematous cornea, pseudohypopyon inferiorly covering 1/3 of the anterior chamber and cataractous lens.

An ocular ultrasonography (B Scan) had detected intralesional calcification with total retinal detachment (Figure 2). CT orbit and
brain showed the presence of calcified mass at the posterior pole with involvement of optic nerve and sclera. There was no extraocular mass involvement and extension to brain (Figure 3). Ultrasound biomicroscopy was used to examine the extension of the lesion to the anterior segment (Figure 4).

By examination under general anesthesia, this case is classified under Group E based on the International Intraocular Retinoblastoma Classification[4]. The patient was referred to paediatric oncology for joint management. Bone marrow aspirate and lumbar puncture were done. Results were normal and no retinoblastoma cell was detected. The genetic study of mutations in the retinoblastoma gene was negative. Patient was planned for 3 cycles of chemotherapy consisting of carboplatin, vincristine, and etoposide for every 3 weeks prior to enucleation with medpor implant.

Histopathological examination showed endophytic retinoblastoma with multifocal lesions in left eye and the largest lesion was 2 mm × 3 mm × 2 mm. There was invasion to the Bruch’s membrane, choroid and sclera. No optic nerve involved (Figure 5).

Post enucleation, patient continued with the chemotherapy up to 6 cycles without complications. The right eye is normal and he used an external ocular prosthesis, which is well tolerated. After 1 year, repeated CT brain and orbit showed no local or distant metastasis.
Diseases mimicking intraocular inflammation and uveitis are termed as masquerade syndrome which can be benign or malignant[5]. Retinoblastoma is rare but is the most common primary ocular malignancy of childhood. Incidence of retinoblastoma is about one in 15,000–20,000 live births[1]. It usually presents as leukocoria, strabismus, proptosis or decreased vision[3]. Only 1%–3% of retinoblastomas present as intraocular inflammation causing uveitis with presence of tumor cells groups simulating hypopyon[6]. About 80% of cases are diagnosed before the age of 4 years, with a median age at diagnosis of 2 years[7].

This patient presented at 4 years with symptoms of severe uveitis with no obvious proptosis and strabismus. As the patient’s right eye was normal with good vision, he did not present with visual problem. A British study reported the incidence of pediatric uveitis to be 3.15 per 100,000 children up to 5 years of age[8].

One year after initiation of treatment, patient was alive and had no symptoms of local recurrence or distant metastasis. Repeated CT brain and orbit did not reveal any recurrence of tumor.

In conclusion, ophthalmologists should have high suspicion of retinoblastoma in children presenting with uveitis, pseudohypopyon or retinal detachment. Prompt action and treatment can reduce the mortality.

Conflict of interest statement

We declare that we have no conflict of interest.

References

[1] Murphree AL, Samuel MA, Harbour JW. Retinoblastoma. In: Ryan SJ, editor. Retina. St. Louis: Mosby-Year Book Inc; 2006, p. 568.
[2] Shields JA, Augsburger JJ. Current approaches to the diagnosis and management of retinoblastoma. Surv Ophthalmol 1981; 25: 347-72
[3] Chuluunbati T, Jamiyannav B, Mankhuu B, Bazarsad U, Molom A, Kao LY, et al. Retinoblastoma in Mongolia: clinical characteristics and survival from 1987 to 2014. Taiwan J Ophthalmol 2016; 6(2): 79-84.
[4] Linn Murphree A. Intraocular retinoblastoma: the case for a new group classification. Ophthalmol Clin North Am 2005; 18: 41-53.
[5] Kubicka-Trzaska A, Romanowska-Dixon B. Malignant uveitis masquerade syndromes. Klin Oczna 2008; 110: 199-202.
[6] Catalá-Mora J, Parareda-Salles A, Vicuña-Muñoz CG, Medina-Zurinaga M, Prat-Bartomeu J. [Uveitis masquerade syndrome as a presenting form of diffuse retinoblastoma]. Arch Soc Esp Oftalmol 2009; 84: 477-80. Spanish.
[7] de Aguirre Neto JC, Antoneli CB, Ribeiro KB, Castilho MS, Novaes PE, Chojniak MM. Retinoblastoma in children older than 5 years of age. Pediatr Blood Cancer 2007; 48(3): 292-5.
[8] Edelsten C, Reddy MA, Stanford MR, Graham EM. Visual loss associated with pediatric uveitis in English primary and referral centers. Am J Ophthalmol 2003; 135: 676-80.
[9] Ghassemi F, Bazvand F, Makateb A. Lesions simulating retinoblastoma at a tertiary care center. J Ophthalmic Vis Res 2015; 10: 316-9.
[10] Krzemień W, Wojcieszak J, Zawilska JB. [Retinoblastoma: genetic background, modern diagnostic methods and therapies]. Przegl Lek 2015; 72(7): 358-65. Polish.
[11] Kachewar SG, Kulkarni MM. A review of limbal epibulbar lesions. J Clin Diagn Res 2014; 8(1): 203-5.
[12] Chung EM, Specht CS, Schroeder JW. From the archives of the AFIP: pediatric orbit tumors and tumoralike lesions: neuroepithelial lesions of the ocular globe and optic nerve. Radiographics 2007; 27(4): 1159-86.
[13] Galuzzi P, Hadjistilianou T, Cerase A, De Francesco S, Toti P, Venturi C. Is CT still useful in the study protocol of retinoblastoma? AJNR Am J Neuroradiol 2009; 30(9): 1760-5.
[14] Dimaras H. Retinoblastoma genetics in India: from research to implementation. Indian J Ophthalmol 2015; 63: 219-26.
[15] Jagadeesan M, Khetan V, Mallipatna A. Genetic perspective of retinoblastoma: from present to future. Indian J Ophthalmol 2016; 64: 332-6.