Journal of Coastal Life Medicine

journal homepage: www.jclmm.com

Case report doi: 10.12980/jclm.4.2016J6-151

Masquerade syndrome: Retinoblastoma presenting as phacolytic glaucoma

Li-Jing Peng[1*], Shuaibah Abdul Ghani[2], May May Choo[1], Syed Shoeb Ahmad[2], Nor Haizura Bt Abd Rani[3]

[1]*Department of Ophthalmology, University of Malaya, Kuala Lumpur 50603, Malaysia*

[2]*Ophthalmology Department, Queen Elizabeth Hospital, Kota Kinabalu 88586, Malaysia*

[3]*Department of Pathology, Queen Elizabeth Hospital, Kota Kinabalu 88586, Malaysia*

ARTICLE INFO

Article history:
Received 17 Aug 2016
Received in revised form 10 Sep 2016
Accepted 15 Sep 2016
Available online 20 Sep 2016

Keywords:
Retinoblastoma
Masquerade syndrome
Leukocoria
Enucleation

ABSTRACT

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.

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. Both eyes were orthophoria. There was no obvious proptosis and nystagmus, and extraocular movement was normal. The conjunctiva of left eye was injected and cornea was oedematous. There was pseudohypopyon inferiorly. Lens was opaque and there was no fundus view. Intraocular pressure was 50 mmHg.

An ocular ultrasonography (B Scan) had detected intralesional calcification with total retinal detachment. CT orbit and brain

*Corresponding author: Jing-Peng Li, Department of Ophthalmology, University of Malaya, Kuala Lumpur 50603, Malaysia.
Tel: +60123286810
E-mail: penglijing85@gmail.com
The journal implements double-blind peer review practiced by specially invited international editorial board members.

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. Ultrasound biomicroscopy was used to examine the extension of the lesion to the anterior segment.

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

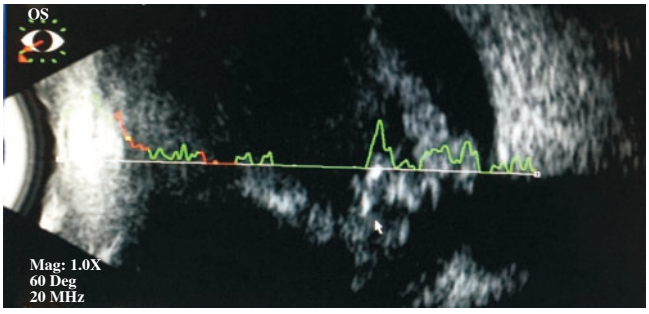

Figure 2. B-scan showing total retinal detachment with intralesional calcium.

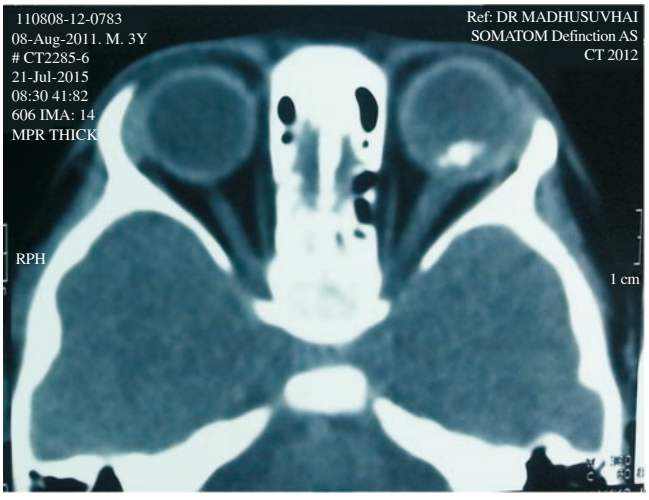

Figure 3. CT orbit showing enhancing lesion with calcification at the posterior pole involving the optic nerve of the left eye and thickening of sclera.

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.

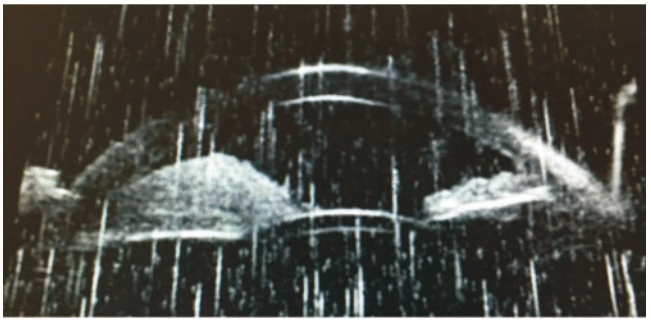

Figure 4. Ultrasound biomicroscopy showing the mass extending till the anterior segment.

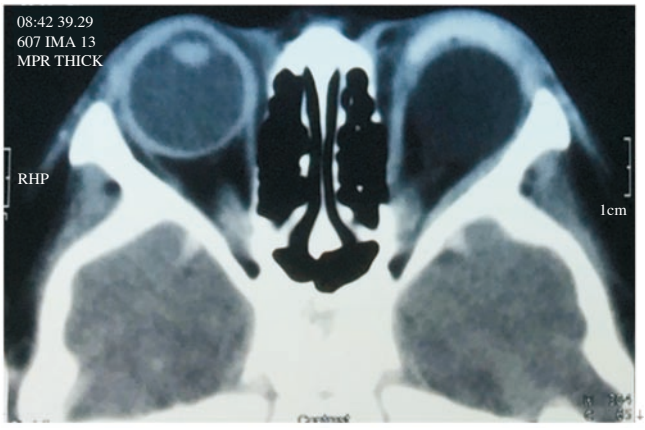

Figure 6. CT orbit after 1 year of diagnosis.

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.

Post enucleation, patient continued with the chemotherapy up to

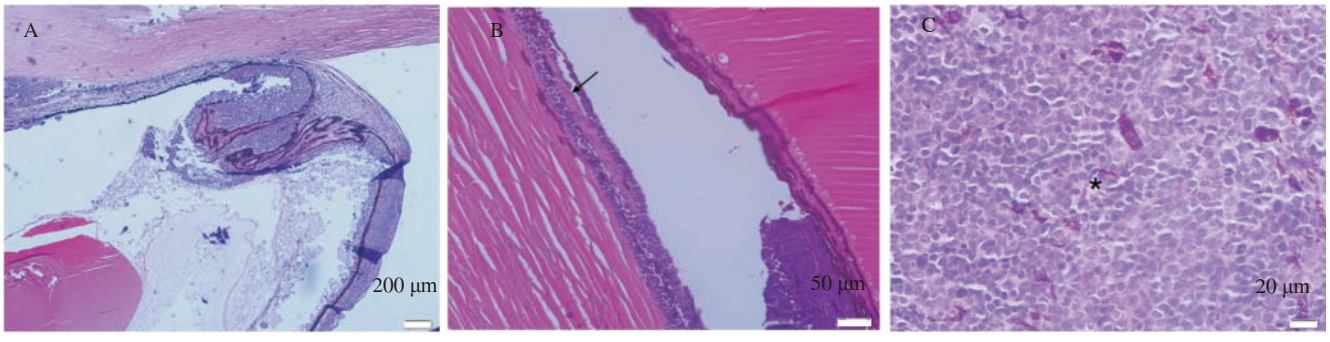

Figure 5. The tumour involved ciliary body and anterior chamber (A), tumour invaded the sclera (arrow) (B), tumour with rosettes formation (*) (C).

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.

3. Discussion

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 15000–20000 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 100000 children up to 5 years of age[8]. Thus, the diagnosis of retinoblastoma should be excluded, as uveitis in the pediatric population is less common than adults. Lesions with clinical similarities to retinoblastoma should also be excluded, as accurate diagnosis is essential for proper management[9]. Diagnosis of retinoblastoma requires an indirect ophthalmoscopic examination. In addition, imaging techniques such as ultrasonography, magnetic resonance imaging and CT are used[10,11].

About 95% of retinoblastoma is calcified[12]. For typical retinoblastoma, it presents as irregular mass with high reflectivity due to calcification in B scan. However, sonographic capacity for detecting small calcified masses is limited by the presence of complex intraocular interfaces associated with vitreous opacities, retinal masses, subretinal fluid and retinal detachment[13]. In this case, ultrasonography and CT orbit confirmed the diagnosis by showing localised mass with calcification.

He was started on chemotherapy consisting of vincristine, carboplatin and etoposide every 3 weeks for 3 cycles. Three weeks later, enucleation of left eye was performed. After the enucleation, he had another 3 cycles of chemotherapy, which was totally 6 cycles of chemotherapy.

Individuals with non-hereditary retinoblastoma have normal *RB1* genes at the germline level and these tumors are always unilateral and unifocal[14]. Histopathological examination post enucleation for this patient showed multifocal lesion and the *RB* gene mutation was negative for this patient. Genetic counseling was given to the patient's family members[15].

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] Chuluunbat T, Jamiyanjav B, Munkhuu 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] Català-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 DS. An imaging review of intra-ocular calcifications. *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 tumorlike lesions: neuroepithelial lesions of the ocular globe and optic nerve. *Radiographics* 2007; **27**(4): 1159-86.

[13] Galluzzi 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.