Content available at: https://www.ipinnovative.com/open-access-journals

IP International Journal of Ocular Oncology and Oculoplasty

Journal homepage: https://ijooo.org/

Case Report

Late-onset retinoblastoma camouflaged with proven chronic uveitis: A case report

Dhivya Ashok Kumar[1],*, Heba Thajudeen[1]

[1] *Dept. of Oculoplasty, Orbit, Oncology, Dr. Agarwal Eye Hospital and Eye Research Centre, Chennai, Tamil Nadu, India*

ARTICLE INFO

Article history:
Received 06-01-2022
Accepted 29-01-2022
Available online 09-05-2022

Keywords:
Retinoblastoma
Late onset retinoblastoma
Chronic uveitis
Panuveitis
Painful blind eye

ABSTRACT

A rare case of a 16year old female, diagnosed elsewhere as chronic uveitis in the left eye positive antinuclear antibodies and Anti-SSA antibodies, treated with steroids and referred for cyclocryotherapy for a painful blind eye. On examination, the patient denied perception of light. There was proptosis, limitation of extraocular movements and corneal melting with no view of the posterior segment. Ultrasound B scan showed dense vitritis with doubtful specks of calcification. Magnetic resonance imaging revealed a soft tissue mass obliterating the vitreous and anterior chamber with retroscleral extension and involvement of the optic nerve. Extended enucleation was performed under General anaesthesia. Histopathological examination confirmed a well differentiated retinoblastoma with extrascleral and optic nerve involvement. Metastatic work up was normal. Intravenous Chemotherapy and post surgery radiotherapy to the socket was advised for further management. Late onset retinoblastoma mimicking ocular inflammation with positive antibody serology is rare and the report shows the need for imaging in refractory uveitis.

1. Introduction

Retinoblastoma (RB) is the most frequently encountered intra-ocular malignancy in children.[1] The incidence is reported to be 1 in 15000 to 18000 live births, particularly diagnosed by 5 years of age.[2–4] Occurrence of this tumour in adults is extremely rare, with few isolated cases reported in the literature.[5] In this report, we discuss a rare presentation of late-onset retinoblastoma in a 16 years old female, misdiagnosed as chronic uveitis, where imaging modalities helped in correcting diagnosis and elective planning. The case report followed the tenets of the declaration of Helsinki and informed consent has been obtained.

2. Case Presentation

A 16 years old female was referred for cyclocryotherapy for a painful blind left eye. Parents gave a history of diminution of vision, associated with redness and pain in the left eye since1year. There was no history of trauma, but there was increasing prominence and later protrusion of the left eye with aggravation of symptoms inspite of treatment. It was reported that the patient had squinting in the same eye since 5years of age, but no ophthalmic evaluation was ever done until the present onset of symptoms. She was diagnosed with chronic uveitis elsewhere and treated with steroids. There was no family history of tumours. Examination of the right eye was unremarkable with 20/20 vision.

On examination, the patient denied perception of light in the left eye. There was dystopia with proptosis and limitation of extra-ocular movements. Presence of lid edema, conjunctival and ciliary congestion, prominent episcleral veins, cataractous lens, exudates in shallow anterior chamber, intercalary staphyloma and

* *Corresponding author*.
E-mail address: susruta2002@gmail.com (D. A. Kumar).

https://doi.org/10.18231/j.ijooo.2022.013

neovascularisation of iris were noted (Figure 1). Corneal melting hindered the view of the posterior segment.

A preliminary Ultrasonogram (USG) Bscan performed showed dense vitritis with few hyperechoic lesions, suspicious of calcification. Magnetic resonance imaging (MRI) showed a T2 hypointense lesion obliterating vitreous and anterior chamber along with a retroscleral extension, associated with irregularity of the posterior sclera and involvement of optic nerve.(Figure 2) A differential diagnoses of either intra-ocular mass lesion or infective etiology, with extra-ocular/episcleral invasion and involvement of the optic nerve was made.

A dilemma over diagnosis prevailed as the clinical signs of a possible infectious etiology did not equate to the symptoms of the patient. The patient had complicated cataract and no proptosis at the time of first presentation (as per old records) which obscured view of the posterior segment. Signs suggestive of uveitis were strengthened by positive anti-nuclear antibody (ANA). However, doubtful calcification seen on USG along with the rapid progression of proptosis and unresponsiveness to treatment weighed more in favour of a possible missed masquerade. MRI image of the homogenous retroscleral involvement surrounding the globe contour also raised the bar of suspicion towards a mass lesion.

Weighing all possibilities, it was decided to perform an extended enucleation under General anaesthesia for the left eye with the informed consent of nil visual prognosis. The surgery was performed and the specimen was sent for histopathological examination (HPE). Post enucleation, a conformer was placed and the patient was given intravenous and topical antibiotics.

HPE confirmed a well differentiated retinoblastoma (Figure 3), with many Flexner-Wintersteiner rosettes, Homer-Wright rosettes, extensive areas of necrosis, extrascleral and optic nerve margin involvement. Patient was classified as Stage 3, group E (International retinoblastoma classification) and systemic work up was negative for metastasis. She underwent chemotherapy with 12 cycles of VEC regimen (Vincristine, Etoposide and Carboplatin) with radiotherapy to the orbit. No recurrence at 1 year follow up.

3. Discussion

Retinoblastoma, a primitive neuro-ectodermal intraocular malignancy, originates from the sensory retina and is usually diagnosed before 5years of age. Only 8.5% of patients are older than this age at diagnosis and the occurrence is almost always sporadic.[6] There is no gender predilection and all previously reported cases of adult onset RB were unilateral.[7]

Theoretically, two hypotheses explain the late presentation; a de-novo occurrence or a malignant transformation of retinocytoma. In the former, there is no

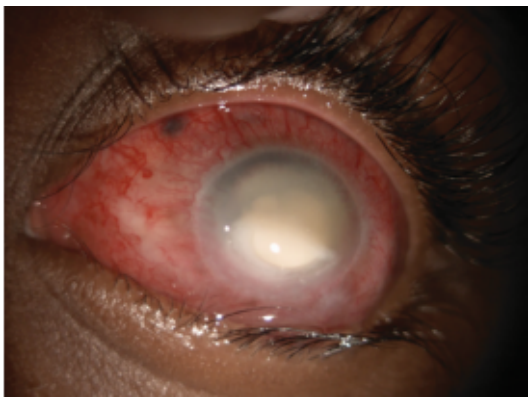

Fig. 1: Clinical picture of left eye of the patient showing corneal melting with exudates in anterior chamber with ciliary congestion and cataractous lens

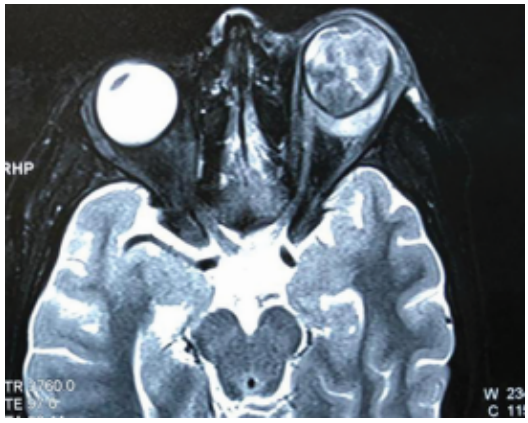

Fig. 2: Magnetic resonance imaging of the orbit showing vitreous echoes with retroscleral thickening with proptosis of left eye

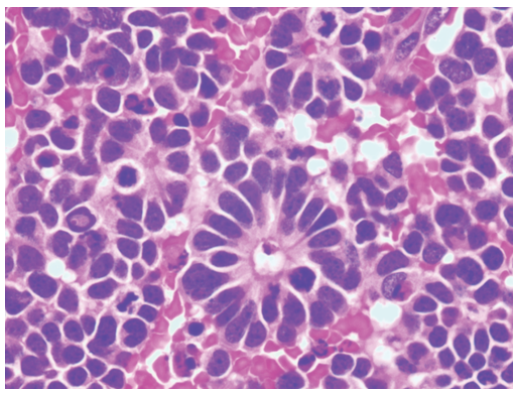

Fig. 3: Histopathology in Hematoxycillin and eosin stain, 10X magnification showing well differentiated retinoblastoma cells showing Flexner-Wintersteiner rosettes

antecedent lesion but a late malignant transformation of rare embryonal retinal cells occurs, whereas in the latter, a previously undiagnosed or spontaneously arrested/regressed RB is present (retinocytoma) which gets reactivated later in life by an additional oncogenic mutation.[8,9] The difficulty in diagnosis stems from the low frequency of occurrence and the atypical presentation.[7] Perhaps, a view of a dome shaped mass in fundoscopy might aid the diagnosis, but often posterior segment view is precluded which makes the diagnosis rely on imaging modalities like USG, computerized tomography or MRI.[10]

It is advocated that any atypical uveitis in older children, may be the first presentation of retinoblastoma, particularly if associated with glaucoma or with poor response to therapy or persistent.[11] In this case, the patient was a young female, advised rheumatological workup in view of a simulated panuveitis. Presence of squint since early childhood with reportedly no visual complaints and initial presentation of uveitis coupled with positivity for auto-antibodies, paved way for a misdiagnosis. We hypothesize that a possible masquerade was thus missed until the advent of a rapidly progressing proptosis.

Atypical uveitis is a known manifestation of RB, especially in older children. But this case is a rare entity because in an otherwise proven chronic uveitis, the possibility of a masquerade/late onset RB did not surface until proptosis set in. We would like to highlight the importance of imaging modalities in diagnostics and that rare chances of a lingering malignancy should be kept in mind especially if the imaging is inconclusive or patient is unresponsive to treatment.

4. Conflict of Interest

The authors declare that there are no conflicts of interest in this paper.

5. Source of Funding

None.

References

1. Petersen R. Retinoblastoma: Diagnosis and nonradiation therapies. Principles and Practice of Ophthalmology. Philadelphia: WB Saunders Co; 1994. p. 3279–82.
2. Bishop JO, Madsen EC. Retinoblastoma: Review of current status. *Surv Opthalmol*. 1975;19(6):342–66.
3. Rubenfeld M, Abramson DH, Ellsworth RM, Kitchin FD. Unilateral vs. bilateral retinoblastoma: correlations between age at diagnosis and stage of ocular disease. *Ophthalmology*. 1986;93(8):1016–9.
4. Maurya RP, Gupta A, Singh V, Gupta V, Singh VP. Pattern of presentation of retinoblastoma in North India: A teaching hospital survey. *J Scientific Res*. 2022;66(1):186–92.
5. Garg V, Gupta A, Pruthi SK, Khare P. Retinoblastoma in an adult. *J*. 2018;35(2):120–4.
6. Shields JA, Shields CL, Ehya H, Jr RE, De Potter P, et al. Fine-needle aspiration biopsy of suspected intraocular tumors: the 1992 Urwick lecture. *Ophthalmology*. 1993;100(11):1677–84. doi:10.1016/s0161-6420(93)31418-1.
7. Sengupta S, Pan U, Khetan V. Adult onset retinoblastoma. *Indian J Ophthalmol*. 2016;64(7):485–91. doi:10.4103/0301-4738.190099.
8. Neronova-Kotova LA. Case of retinoblastoma in an adult. *Vestn Oftalmol*. 1986;102:75.
9. Kremlicka L, Roubková H. Retinoblastoma in an adult (author's transl). *Klin Monbl Augenheilkd*. 1975;167(1):119–23.
10. Raj A, Arya SK, Punia RS, Kohli P. Adult onset retinoblastoma: a diagnostic dilemma. *Orbit*. 2016;35(1):51–3.
11. Richards WW. Retinoblastoma simulating uveitis. *Am J Ophthalmol*. 1968;65(3):427–31.

Author biography

Dhivya Ashok Kumar, Consultant

Heba Thajudeen, Resident

Cite this article: Kumar DA, Thajudeen H. Late-onset retinoblastoma camouflaged with proven chronic uveitis: A case report. *IP Int J Ocul Oncol Oculoplasty* 2022;8(1):61-63.