

CASE REPORT

RETINOBLASTOMA IN AN OLDER CHILD: A CASE REPORT

Kumudini Subedi^{1*}, Prachi Dulal¹, Srijana Parajuli¹, Sangita Pradhan¹

¹Department of Uveitis and Vitreoretinal surgery, Bharatpur Eye Hospital, Bharatpur, Chitwan, Nepal

Received: 7 Jan, 2024

Accepted: 11 Mar, 2024

Published: 30 Mar, 2024

Key words: Biopsy; Children; Malignancy; Retinoblastoma.

**Correspondence to:* Kumudini Subedi, Department of Uveitis and Vitreoretinal surgery, Bharatpur Eye Hospital, Bharatpur, Chitwan, Nepal.
Email: kumusubedi@gmail.com

DOI: <https://doi.org/10.54530/jcmc.1466>

Citation

Subedi K, Dulal P, Parajuli S, Pradhan S. Retinoblastoma in an older child: a case report. Journal of Chitwan Medical College. 2024;14(47):95-7.



Peer Reviewed

ABSTRACT

Retinoblastoma is the most common intraocular malignancy of childhood. More than 90% of the patients are under 5-years old. It is uncommon in age above 5 years. In this study we report a case of retinoblastoma in a 9-year-old girl. She presented to our department with decreased vision in left eye noted for 6 days. Vision loss was non-progressive, associated with mild pain. Snellen's acuity in the affected eye was light perception only. There were no signs of active inflammation in the anterior chamber. Anterior vitreous face demonstrated exuberant vitreous inflammation. A large, elevated, sessile lesion occupying the inferotemporal quadrant was seen just behind the lens with significant adjacent inflammation. Right eye examination was unremarkable. Ultrasound and Magnetic Resonance Imaging demonstrated retinoblastoma with no extra-ocular spread. Enucleation was done and biopsy of ocular tissue and bone marrow trephine biopsy was found normal. We conclude that any suspicious lesion in childhood must be evaluated for retinoblastoma. Even in older child the retinoblastoma may be localized and may have good visual prognosis.

INTRODUCTION

Retinoblastoma, is well known as the most common intraocular malignancy in children under 5 years of age. Less than 10% of diagnosed cases of retinoblastoma are older than 5 years owing to its rarity, lower clinical suspicion and uncommon clinical presentation.^{1,2} The incidence of unilateral cases is higher than bilateral cases as the age increases with approximately 98% bilateral cases being diagnosed before 3 years of age.³ Retinoblastoma in older children presents with clinical features uncommon to the classical clinical features of leukocoria and strabismus in young patients which often leads to misdiagnosis or late diagnosis.⁴ Thus, in older children the disease is often aggressive with extraocular and /or systemic involvement at presentation needing enucleation with or without adjuvant chemotherapy or radiation.^{4,5}

We report a case of unilateral retinoblastoma in a 9-year-old female. Retinoblastoma in an older child is known to forecast grave ocular and systemic prognosis. However, early diagnosis and prompt treatment in our case led to prevention of possible extraocular and systemic spread. This is a rare presentation which had the potential of life-threatening outcome if precise

management couldn't be done. Thus, any unexplained vision loss and suspicious ocular inflammatory condition should be investigated for retinoblastoma irrespective to the age of the child.

CASE REPORT

A 9-year-old girl presented to outpatient department (OPD) of Bharatpur Eye Hospital (BEH) with complaint of noticeable vision loss in left eye (LE) for 6 days. The vision loss was associated with eye ache, redness and white reflex in the same eye. There was no history of similar episode in the same or fellow eye. There was no history of similar disease in her siblings. The only relevant systemic history was history of appendectomy done 3 months ago. Surgery apparently had uneventful post-operative course and there were no postoperative complications. Uncorrected Snellen's visual acuity was 6/6 in right eye (RE) and perception of light with projection of rays of light inaccurate in all quadrants in LE with no improvement observed with pinhole and/or refraction. Further examination of anterior segment of LE revealed a sluggishly reactive pupil, clear cornea with deep anterior chamber and cellular reaction of 3+ by SUN classification. The lens was clear, in proper anatomical position and anterior

vitreous face demonstrated cells >50 cells per high power field. Posterior segment evaluation under mydriatics revealed vitreous clumps, vitreous snowballs and whitish elevated lesion on the inferior-temporal quadrant grossly from 3 to 5 o'clock position as seen in the anterior vitreous face with surface telangiectatic vessels. Optic nerve head was not discernable amidst dense vitreous clumps. RE was unremarkable. USG B scan revealed hyperechoic retinochoroidal mass filling almost 2/3rd of the vitreous cavity with high surface reflectivity and internal reflectivity and variable shadowing due to areas of calcification and necrosis (Figure 1).

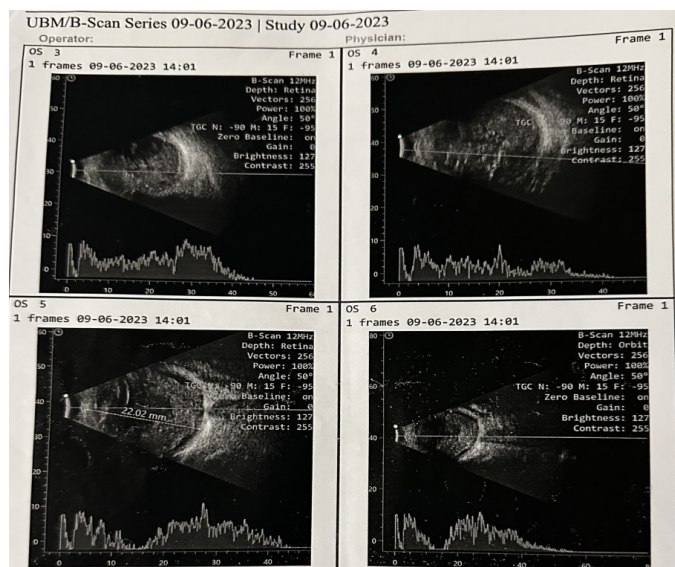


Figure 1: USG B scan LE revealing hyperechoic retinochoroidal mass

MRI brain and orbit was done to confirm the diagnosis and also to determine extra-ocular spread of tumor. MRI brain and orbit revealed large lobulated mass in the inferior quadrant of the LE with intermediate signal in the T1W1 and hypo-intense signal in T2W1 suggestive of retinoblastoma. No extra-ocular extension was noted. Brain parenchyma was normal in signal intensity with no focal or diffuse mass lesion (Figure 2A, 2B, 2C).

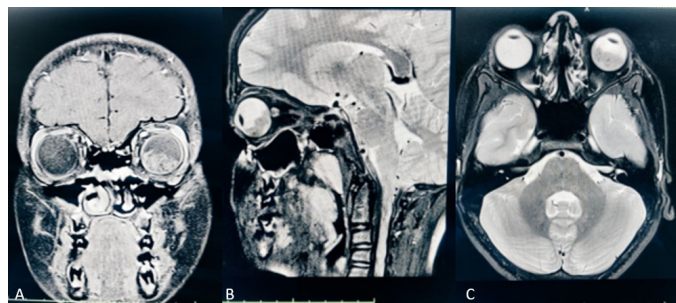


Figure 2: MRI Brain and orbit A. Coronal View B. Sagittal View C. Axial View

A provisional diagnosis of endophytic retinoblastoma of LE was made. Patients' father was counseled who was skeptical and sought opinion at other centers especially regarding the need for post-operative chemotherapy. Finally, patient underwent examination under anesthesia of both eyes with LE enucleation

with ball implant and conformer placement and temporary tarsorrhaphy at another center in India. Patient also underwent CSF study and bone marrow biopsy in the same setting to rule out any systemic spread or syndromic association. Left eye enucleation sample was sent for histopathology. Genetic testing was also done which did not suggest any suspicious genotype. There was no lesion suggestive of retinoblastoma in fellow eye. Histopathology revealed endophytic growth pattern of the posterior segment with necrosis and calcification and 80% viable cell. The tumor cells were well differentiated with no severe anaplasia. There was > 3mm (around 30%) invasion of choroid. Adjacent scleral invasion was seen of partial thickness. Invasion of prelaminar and laminar portion of the optic nerve was noted. The anterior segment was free from invasion. Pathologic classification of the primary tumor (T) –pT2b was made. CSF analysis and bone marrow aspirate was negative for retinoblastoma cells. The tumor was classified as Stage I N1 according to International Retinoblastoma Staging System. After 6 weeks of enucleation, temporary tarsorrhaphy suture was removed and ocular prosthesis placement was done as wound healing was confirmed.

DISCUSSION

Retinoblastoma in children more than 5 years of age is a rare presentation as shown by mean age-adjusted annual incidence of 11.8 per million for children aged 0–4 years and 0.6 per million in children aged 5 years and older.³ Consequently children over 5 years of age constitute only 3.5% to 8.5% of all retinoblastoma cases.¹⁻³ Previous case series published from Nepal with a gross figure of 130 cases in total state that only 2 cases were over 8 years of age.⁴⁻⁸ An extremely rare case reported from Nepal stated retinoblastoma to have occurred in a 37 year old male.⁸ The pathophysiology for late onset retinoblastoma is postulated to be the malignant transformation of benign retinoma (retinocytoma) or RB1 mutation in persistent embryonic retinal cells.⁹ The presence of clinical features unusual to that of younger children and delay in suspicion and referral due to its rare occurrence in that particular age group further leads to delay in diagnosis and treatment.² Various studies have shown atypical clinical features including decreased vision, floaters, masquerade syndrome, vitreous hemorrhage as more common in older retinoblastoma cases compared to the stereotype description of leucocoria and strabismus in younger children.^{9,10} This leads the treating clinician towards other differentials like coats disease, uveitis, endophthalmitis, toxocariasis and vitreous hemorrhage with retinal detachment.^{9,10} Meel R et. al. studied clinical and histopathological features and treatment outcomes of retinoblastoma cases presenting at an older age and concluded that a significant percentage of retinoblastoma in developing countries is misdiagnosed (upto one-third of cases) and may present at an advanced stage in 46% of cases.² The delay in diagnosis increases risk of extra-ocular spread and leads to higher mortality. Studies have reported poor prognosis in cases of retinoblastoma in older children.¹¹ In the present case, the child had an acute presentation with atypical signs and symptoms with no involvement of the fellow eye or family history. Early diagnosis and prompt management of the patient

led to prevention of further extra-ocular extension and systemic metastasis. This also helped in boosting the confidence level of the child and psycho-social rehabilitation.

CONCLUSION

Retinoblastoma is rare in older children and there is a high

likelihood of misdiagnosis. Consequent delayed referral may lead to not only poorer visual and anatomical outcomes but also psycho-social impairments and higher mortality rates. Thus, retinoblastoma must be considered in the differential diagnosis even when an older child presents in the eye care facility if there are suggestive or suspicious ocular findings.

REFERENCES:

1. Cebeci Z, Tuncer S, Kebudi R. Clinical features and long-term follow-up of patients with retinoblastoma in Turkish children older than 5 years of age. *Journal of Ophthalmology*. 2020 Jan 20;2020. [\[DOI\]](#)
2. Meel R, Kashyap S, Bakhshi S, Singh Bajaj M, Wadhvani M. Retinoblastoma in children older than 6 Years of age. *Ocular oncology and pathology*. 2020 Dec 15;6(6):395-404. [\[DOI\]](#)
3. Broaddus E, Topham A, Singh AD. Incidence of retinoblastoma in the United States: 1975-2004. *British Journal of Ophthalmology*. 2008 Jul 11. [\[DOI\]](#)
4. Limbu B, Kafle PA, Lamichanne DH, Pant SR, Shakya S, Rasaily SB. Epidemiological and Clinical Presentation of Retinoblastoma among Nepalese Children in 2019. *South Asian Journal of Cancer*. 2022 Nov 2. [\[DOI\]](#)
5. Lavaju P, Badhu BP, Shah S, Chaudhary SK, Upadhyaya P. Clinical profile of retinoblastoma presenting in a tertiary care hospital, eastern Nepal. *Health Renaissance*. 2015;13(3):129-36. [\[DOI\]](#)
6. Saiju R, Thakur J, Karmacharya PC, Shah DN. Retinoblastoma in Nepal: a clinical profile of 30 cases. *Nepal Medical College Journal: NMCJ*. 2006 Sep 1;8(3):171-5. [\[PMID\]](#)
7. Saiju R, Moore G, Shrestha U, Shrestha MK, Ruit S. Retinoblastoma: geographic distribution and presentation at a tertiary eye care centre in Kathmandu, Nepal. *Nepalese Journal of Ophthalmology: a Biannual Peer-reviewed Academic Journal of the Nepal Ophthalmic Society: NEPJOPH*. 2013 Jul 1;5(2):169-76. [\[DOI\]](#)
8. Shrestha A, Adhikari RC, Saiju R. Retinoblastoma in a 37 years old man in Nepal: a case report. *Kathmandu University Medical Journal (KUMJ)*. 2010 Apr 1;8(30):247-50. [\[DOI\]](#)
9. Masoomian B, Dalvin LA, Michael DY, Stathopoulos C, Shields CL. Retinoblastoma in older patients: a retrospective comparative analysis of 100 consecutive patients based on age. *Saudi Journal of Ophthalmology*. 2019 Jul 1;33(3):243-50. [\[DOI\]](#)
10. Shields CL, Shields JA, Shah P. Retinoblastoma in older children. *Ophthalmology*. 1991 Mar 1;98(3):395-9. [\[DOI\]](#)
11. Karcioglu ZA, Abboud EB, Al-Mesfer SA, Al-Rashed W, Pilapil DH. Retinoblastoma in older children. *Journal of American Association for Pediatric Ophthalmology and Strabismus*. 2002 Feb 1;6(1):26-32. [\[DOI\]](#)