UNILATERAL SPORADIC RETINOBLASTOMA IN AN 11-YEAR-OLD CHILD: A RARE CASE REPORT

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ABSTRACT

BACKGROUND

Retinoblastoma is the commonest intraocular tumour of childhood. Majority of cases are diagnosed before 5 years of age, with late presentation being a rarity.

CASE REPORT

An 11-year-old boy presented to our OPD with diminution of vision and white pupillary reflex in the left eye since 1 month. Right eye was normal. Fundus examination of left eye revealed a mass in the inferior quadrant with vitreous haemorrhage. B-scan and MRI were suggestive of retinoblastoma and showed no signs of optic nerve involvement. Subsequently, the patient underwent enucleation of left eye. Primary orbital implant was given. Diagnosis was confirmed histopathologically.

CONCLUSION

Late presentation of retinoblastoma though uncommon can cause acute visual impairment. Retinoblastoma should be considered in the differential diagnosis of leukocoria in any age group.

KEYWORDS

Retinoblastoma, B-scan, MRI, Optic Nerve, Enucleation.

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INTRODUCTION: Retinoblastoma is the most common primary intraocular tumour in children and results from mutations in the tumour suppressor retinoblastoma gene (RB1) located in chromosome 13.[1] It is thought that retinoblastoma arises from retinoblasts, which are retinal precursor cells.[2] The current incidence ranging from one in 15,000 to one in 20,000 live births.[3] The average age when children are found to have retinoblastoma is 9 to 12 months in bilateral cases and 24 months in unilateral cases.[4] It is bilateral in about 25-35% of cases.[5] Out of the unilateral cases, approximately 15% are caused by germinal mutations while the 85% are sporadic.[6]

CASE REPORT: A 11-year-old boy presented to our OPD on 28 September 2015, with diminution of vision and white pupillary reflex in the left eye [Figure 1] since 1 month. There were no other associated symptoms, no past medical history of note, and no history of trauma or eye infection. Family history was not significant. He was born at term through a normal vaginal delivery and had no postnatal complications.

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Figure 1: Leucocoria in the Left Eye

On examination, right eye was normal. In the left eye, visual acuity was perception of hand movement, anterior segment examination was normal, intraocular pressure was 14 mmHg. In the posterior segment examination, there was a dome-shaped mass with vitreous haemorrhage.

A differential diagnosis of retinoblastoma, Coat's disease and toxocarial granuloma was made.

B-scan ultrasound of the left eye revealed a heterogeneous mass of size 1.4 x 1 cm with calcification noted in the temporal retina projecting into the vitreous with echogenic debris in vitreous [Figure 2]. MRI brain and orbit showed left eyeball intra-ocular globular chorioretinal soft tissue lesion at the posteroinferior and lateral quadrant measuring 15 x 10 mm in size with extra ocular bulging. The lesion was isointense on T1 [Figure 3] and hypointense on T2 images [Figure 4]. No optic nerve involvement was seen. MRI of the brain was normal. Right eyeball appeared normal.

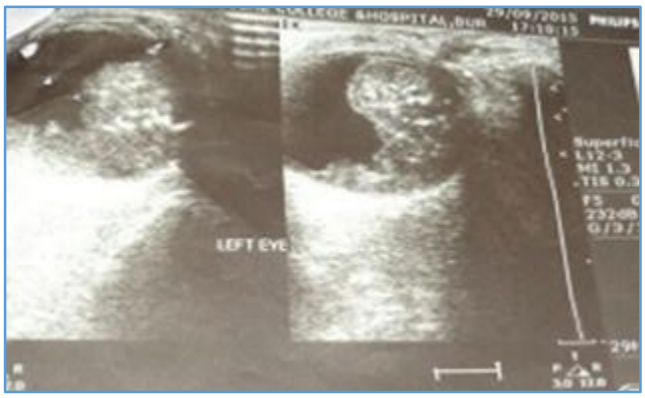

Figure 2: B scan showing Retinal Mass with Focal Vitreous Seeding in the Left Eye

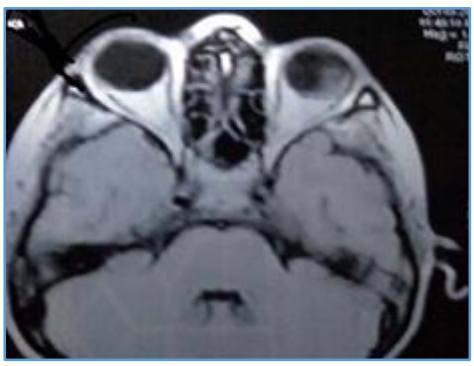

Figure 3: T1 MRI Image Showing Isointense Lesion

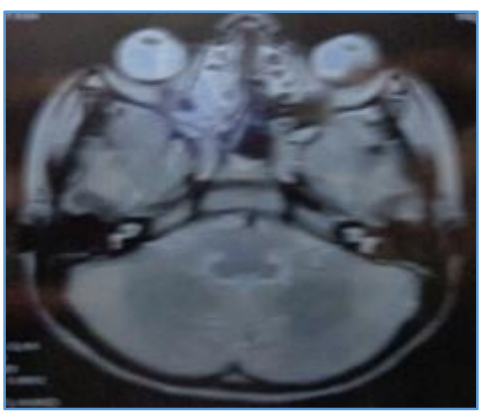

Figure 4: T2 MRI Image Showing Hypointense Lesion

Other investigations including cerebral spinal fluid examination, bone marrow aspirate, toxocara serology, and serum protein studies were normal. Henceforth, a diagnosis of retinoblastoma left eye was made. The tumour was staged as Group C according to the International Classification of Intraocular Retinoblastoma system.

Neoadjuvant chemotherapy was advised. Left eye enucleation was done [Figure 5]. Orbital implant and conformer was given. The procedure was uneventful and the whole eyeball along with the optic nerve stalk was sent for histopathological examination. There were no postoperative complications [Figure 6].

The diagnosis of retinoblastoma was confirmed histopathologically. Macroscopically, a tumour measuring 15 x 10 mm with hypo and hyperpigmented areas and haemorrhagic foci was shown [Figure 7]. Histology revealed atypical proliferation of cellular elements with small round hyperchromatic nuclei and scanty cytoplasm. The neoplastic cells were differentiated in some areas accompanied by Flexner-Wintersteiner rosette formation and mitotic figures [Figure 8]. The choroid and optic nerve were uninvolved.

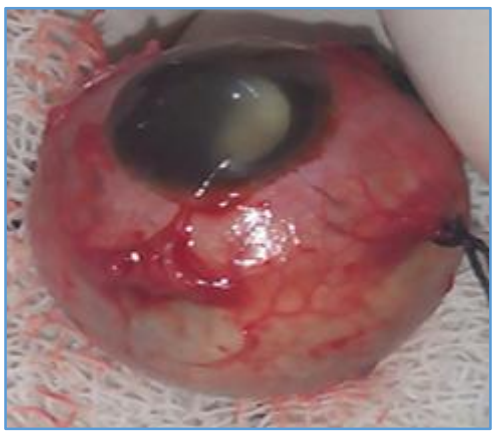

Figure 5: Left Eye Enucleated

Figure 6: Post-operative day 1 Photograph of Patient with Implant and Conformer in situ

Figure 7: Cut section of Enucleated Left Eyeball showing the Tumour Mass with Haemorrhagic FOCI

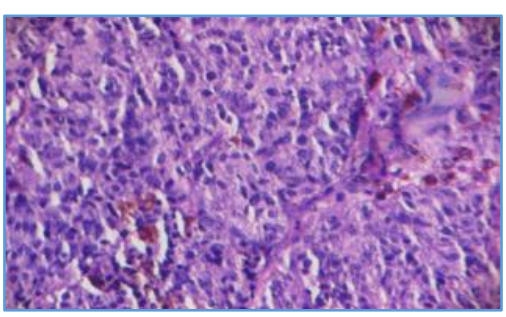

Figure 8: Histopathological Section showing Flexner-Wintersteiner rosettes and Necrotic Areas

DISCUSSION: In majority of cases of retinoblastoma, the diagnosis is made before 5 years of age.[7] According to Shields et al, only 8.5% of patients with retinoblastoma are older than 5 years at the time of diagnosis and all reported cases were sporadic .[8] A study done by Rajasekhar Harini et al. showed that among 81 retinoblastoma patients, 90.12% were sporadic.[9]

Due to its low frequency, retinoblastoma in adults creates a diagnostic dilemma.[10] Although late presenting retinoblastoma is uncommon, there are a number of case reports of these cases.[10, 11, 12, 13] A similar case of late presentation of retinoblastoma in a teen with Aicardi Syndrome has been reported from Houston.[14]

There are a number of explanations for the late presentation of retinoblastomas. Persistence of rare embryonal retinal cells may lead to malignant transformation in later life[15]. The tumour may also arise from previously undiagnosed, spontaneously regressed/arrested retinoblastomas which have been reactivated. There are few reports of retinoblastoma arising from previously documented retinoma or quiescent retinoblastoma[16]. Galli et al suggested a modification in the original mutation model of oncogenesis proposed by Knudson.[17]

If retinoblastoma could be diagnosed in its early stage, less aggressive treatment modalities such as chemotherapy, photocoagulation, cryotherapy, brachytherapy, external beam radiation, and diathermy would be extremely useful.[18] Management of retinoblastoma should be guided by the objectives such as to save life, retain anatomical integrity of the eye, preserve vision, and obtain good cosmetic results.[4] Enucleation is indicated for unilateral retinoblastomas which fill most of the globe and when there is little hope of salvaging vision.[19] Enucleation was the primary treatment modality in the majority of reported late cases of retinoblastoma, as the lesions were detected at a fairly advanced stage and each patient had one normal unaffected eye.

CONCLUSION: Retinoblastoma should be considered in the differential diagnosis of leukocoria at any age group. Successful treatment and prevention of spread of retinoblastoma can be possible with early diagnosis and timely intervention.

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