Case Report on Bilateral Retinoblastoma in Older Children

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Abstract

Retinoblastoma is a rare intraocular malignant tumour more commonly seen in children below five years of age. We presented a rare case of retinoblastoma in a 7 year old child who came to the ophthalmology OPD with complaints of loss of vision in both eye and deviation in right eye. Fundus examination showed multiple floating cotton ball opacities in vitreous cavity. B-scan was suggestive of Group E retinoblastoma in right eye and Group D retinoblastoma in left eye. To conclude, any unexplained visual loss in children of any age should be examined extensively by an ophthalmologist, as the risk of retinoblastoma cannot be ruled out.

Key Words: Retinoblastoma, Leukocoria, Embryonal retinoblasts, Enucleation

Introduction

Retinoblastoma is the most common intraocular tumour in children and is an embryonal malignancy of the retina. It is an autosomal dominant genetic disease caused by a mutation in the RB1 gene on chromosome 13q14. The average age at diagnosis for unilateral cases is 1 year, and for bilateral cases it is 2 years1. It has, however, been identified in older children up to the age of 16. About 40% of retinoblastoma patients have inherited an RB1 gene mutation from their parents. Eighty-five percent of cases are bilateral, ten percent are unilateral, and sixty percent are non-familial (unilateral). Being a tumour suppressor the loss of this gene is thought to result in the development of tumours2,3. Clinically, the tumour most often manifests as strabismus (20%) and leucocoria (60%), with secondary glaucoma, pseudouveitis, orbital inflammation, proptosis, metastatic characteristics, and increased intracranial pressure4.

Material and Method

A 7 year old male child was brought to the ophthalmology OPD by his mother with chief complaints of diminution of vision in both eyes, more in right eye since last 5 months. She specifically described her child not able to read books properly also. Mother also complained that there has occurred deviation of right eye since last 3 months. Patient also complains of dull intermittent and non radiating pain and the right eye being more prominent than the other eye. The symptoms emerged gradually, and there was no history of similar ocular conditions in the family.

- No history of
- Leucocoria [yellowish white papillary reflex]
- Nystagmus
- Redness
- Bleeding from eye

On general examination

- Blood pressure: 120/80 mmHg
- Rest of the general examination was within normal limit.

- On neurological examination patient was fully alert and oriented and had fluent speech. There were no signs of meningeal irritation.

- All cranial nerve examination was normal.

Ophthalmologic Examination

Fundus examination done on INDIRECT OPHTHALMOSCOPY WITH PLUS 20 D LENS
**On anterior and posterior segment evaluation**

<table>
<thead>
<tr>
<th></th>
<th><strong>Right eye</strong></th>
<th><strong>Left eye</strong></th>
</tr>
</thead>
<tbody>
<tr>
<td>Vision</td>
<td>Counting finger 2 mt [with no improvement on pinhole]</td>
<td>Counting finger 3 mt [with no improvement on pinhole]</td>
</tr>
<tr>
<td>Anterior segment</td>
<td>Esotropia 15 degree .</td>
<td>Within normal limit</td>
</tr>
<tr>
<td>Posterior segment</td>
<td>Media Hazy</td>
<td></td>
</tr>
<tr>
<td>Disc</td>
<td>Not able to visualize</td>
<td>Faintly visible , details can not be described</td>
</tr>
<tr>
<td>Blood vessels</td>
<td>Not seen</td>
<td>Fine</td>
</tr>
<tr>
<td></td>
<td>Multiple floating cotton ball opacities floating in vitreous cavity</td>
<td>Multiple floating cotton ball opacities floating in vitreous cavity, suspected irregular mass extending from disc</td>
</tr>
</tbody>
</table>

**B scan** was suggestive of multiple echoes in vitreous cavity with highly echoic mass (calcified) on & around optic nerve area. no evidence of Retinal detachment.

After correlating it clinically diagnosis was made

Right Eye : Group E Retinoblastoma [Extensive retinoblastoma occupying > 50% of the globe with or without neovascular glaucoma, haemorrhage, extension of tumour to optic nerve or anterior chamber].

Left Eye : Group D Retinoblastoma[Tumour with diffuse sub retinal or vitreous seeding > 3mm from tumour].

![Fig. 1. Fundus examination (right eye)](image)
Fig. 2. Fundus examination (left eye)

Fig. 3. B-scan (right eye)
Patient was referred to higher centre where he had undergone enucleation with orbital implant in right eye. Management is still in progress for left eye

**Discussion**

Despite being the most common primary intraocular tumour in the paediatric age group, retinoblastoma remains a rare tumour in children. It is more common in children, with up to 95 percent of cases reported before the age of five. When seen in older children, retinoblastoma is typically unilateral and sporadic. It begins as a single somatic cell that grows into a cancerous tumour. Just 3.9 percent of retinoblastoma was observed in children over the age of 5 in a retrospective analysis conducted by Chang et al. Less than 1% of the 1205 patients had cases between the ages of 7 and 14.

Knudson’s “two-hit” hypothesis states that the carcinogenesis of retinoblastoma is caused by two different mutations in the RB1 tumour suppressor gene. Hereditary cases account for 40% of all cases, while non-hereditary cases account for 60%. Retinoblastoma is a malignant tumour that grows from immature retinal cells called retinoblasts. This ‘two-hit’ oncogenic mutation emerges between the third month after conception and the age of four years, when retinoblasts grow and mature to their maximum potential. This helps to understand why retinoblastoma is almost exclusively present in children under the age of five. Our case is 7 years old, which is considerably above than the average age. However, no further genetic study was carried out in this case to validate the genetic correlation. The causes for retinoblastoma’s late presentation in children over the age of five years are uncertain and contentious. It was assumed that late-onset retinoblastoma may be caused by the persistence of embryonal retinoblasts, or that the tumour may have originated from the reactivation of a previously arrested tumour. Retinoblastoma in older children can have a different clinical appearance than in younger children. The most common symptoms of retinoblastoma in children are leukocoria and strabismus, but older children can also experience reduced vision, eye pain, photophobia, and lacrimation. Uveitis, vitreous opacity, vitreous haemorrhage, retinal detachment, orbital inflammation, or neovascular glaucoma are all rare but probable signs.
Improved diagnostic techniques such as computed tomography (CT), magnetic resonance imaging (MRI) and B-scan certainly help in the diagnosis of these conditions. Where a diagnosis is uncertain, physicians must balance the advantages of enucleation against the risks of not enucleating. Enucleation of one eye would certainly have a significant psychological and social effect on the affected children. On the other hand, failure to diagnose and treat retinoblastoma can result in severe morbidity and even mortality in patients. On a safer side it is still better to enucleate the eye when there is uncertainty in diagnosis of retinoblastoma.

**Conclusion**

Retinoblastoma is a treatable condition that should be treated holistically. Despite the fact that retinoblastoma is more prevalent in younger children, it can also affect older children. Any unexplained visual loss in children, regardless of age, should be referred to an ophthalmologist for a thorough review.

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**References**