

Retinoblastoma Extending up to the Optic Chiasma and Presenting as a Suprasellar Mass

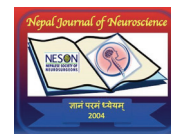
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Abstract

Retinoblastoma is the most common ocular malignancy of childhood. It is present in childhood with leukocoria and strabismus. Most patients are diagnosed under 3 years of age.

Fundoscopic examination can reveal an intraocular mass, but imaging is essential for complete evaluation of the lesion. Although ultrasound is a non-invasive and relatively inexpensive screening tool, cross-sectional imaging is required to assess the involvement of optic nerve and intracranial spread.

We report a case of retinoblastoma in a 5 year old male child who presented with headache diminution of vision in both eyes. Contrast enhanced magnetic resonance imaging (MRI) revealed an enhancing soft tissue mass in the right eye with involvement of optic nerve and optic chiasma and contiguous with a large suprasellar mass causing hydrocephalus. Non-contrast computed tomography (CT) showed extensive calcifications in the mass lesion.

Key words: CT, Imaging, MRI features, Retinoblastoma, Staging.

Introduction

Retinoblastoma is a malignancy of the paediatric age group which arises from the primitive photoreceptors or neuronal retinal cells in the eye. Its global incidence is about 1 in 16,000-18,000 live births, although it may be slightly higher in middle and low income countries.¹

When the disease is unilateral (in around 60% of cases), the median age at diagnosis is 2 years. When the disease is bilateral (in around 40% of cases), the median age at diagnosis is 1 year.² Most patients present with leukocoria and strabismus, although other symptoms may be observed, including rubeosis iridis, buphthalmos, hypopyon, hyphema.²

Classic imaging findings in retinoblastoma include a retrolental mass which often contains foci of calcification. Ultrasound is often used as a preliminary investigation while cross-sectional imaging such as computed tomography (CT) and magnetic resonance imaging (MRI) can confirm the diagnosis by demonstrating an endophytic or exophytic mass which enhances avidly on post-contrast studies.³

Retinoblastomas may spread hematogenously, via the lymphatics, or along the optic nerve sheath. Vitreal seeding and drop metastases within the subarachnoid space are common.^{4,5} Chiasmal involvement which presents as a suprasellar mass lesion is relatively uncommon, which we describe in our case report.

Case report

A 5-year-old male child presented to the out-patient department with complaints of headache and severe diminution of vision in both eyes. Dilated fundoscopy revealed a cream coloured endophytic mass with adjacent dilated blood vessels in the right eye. Fundoscopic examination of the left eye revealed no significant abnormalities.

Contrast enhanced MRI showed an avidly enhancing soft tissue lesion in the posterior segment of the right eye, extending posteriorly along the optic nerve, involving the optic chiasma, and forming a suprasellar mass effacing the third ventricle and causing moderate dilatation of both the lateral ventricles. Optic canal was widened and mild proptosis was present. (Figures 1, 2 and 3)

Screening non-contrast CT scans of the brain and orbits revealed extensive calcification in the intraocular lesion, the optic nerve, and the suprasellar mass. (Figures 4 A, B and C)

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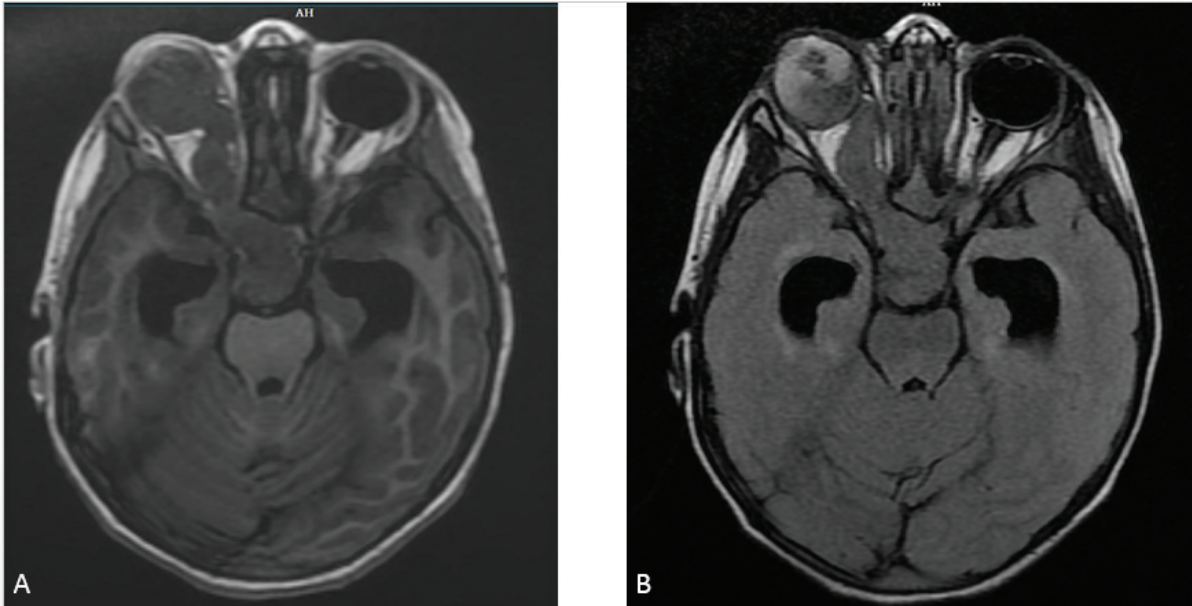


Figure 1: (A) Axial T1 and (B) T2-FLAIR images at the level of the orbits show an isointense soft tissue mass in the right globe suggestive of retinoblastoma. It involves the right optic nerve as well as extending intracranially through the optic canal to involve the optic chiasma.

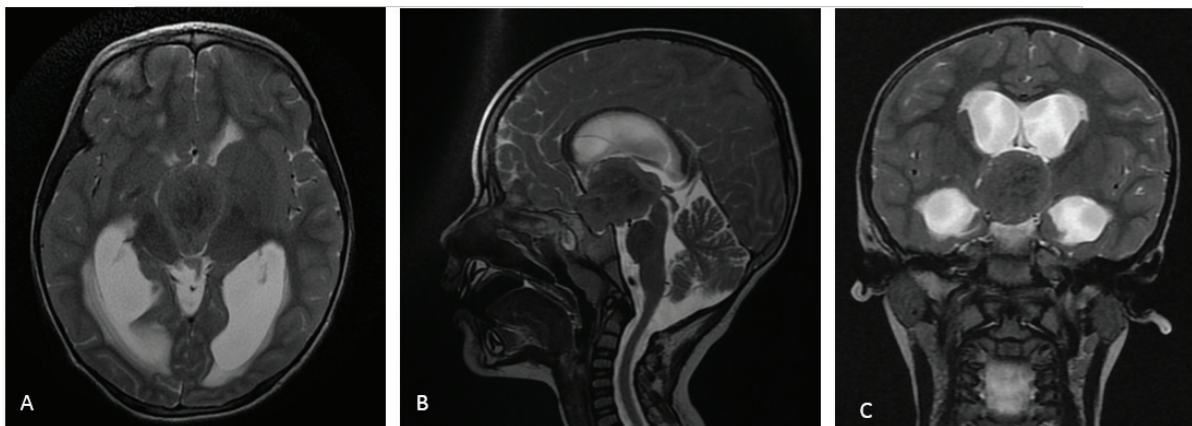


Figure 2: (A) Axial, (B) sagittal, and (C) coronal T2 images showing the true intracranial extent of the retinoblastoma. This suprasellar mass is seen compressing the third ventricle leading to dilatation of the lateral ventricles.

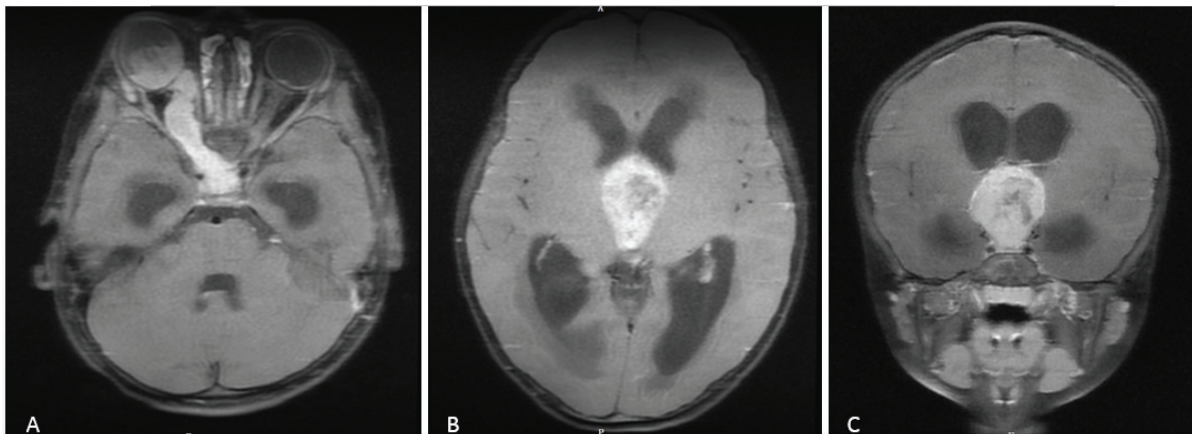


Figure 3 (A, B, C): Post contrast MR images show that the soft tissue component of the retinoblastoma is avidly enhancing.

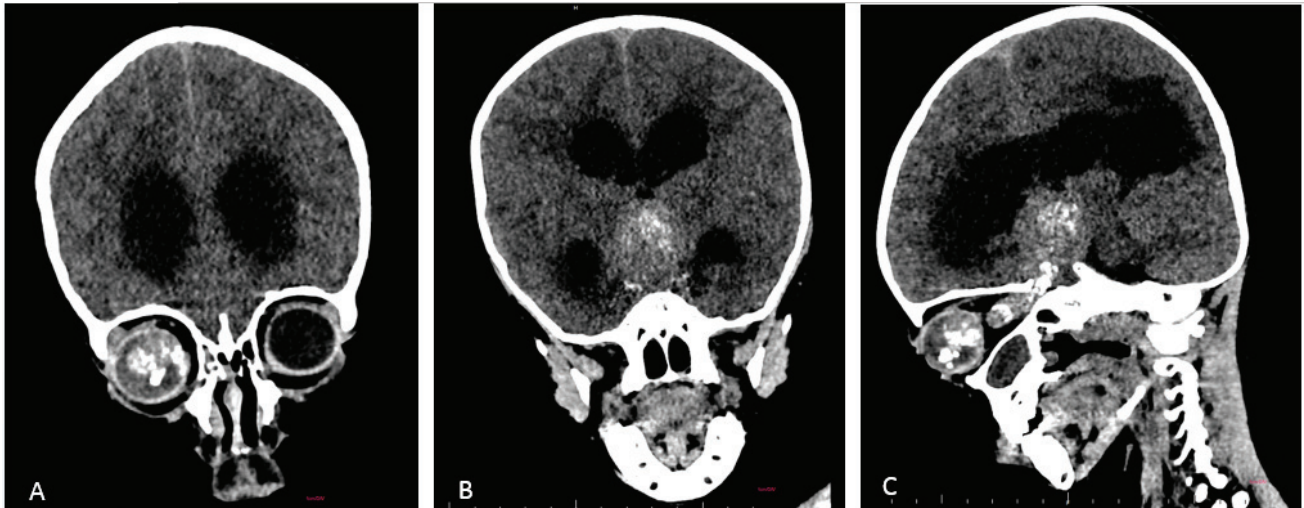


Figure 4 (A, B, C): Non-contrast CT images showing extensive calcification of the retinoblastoma which has involved the optic nerve and optic chiasma, and presented as a suprasellar mass.

Discussion

Retinoblastoma is the most common primary intraocular malignant tumour in the paediatric age group. Around 40% of retinoblastomas are heritable, frequently bilateral, and present early (median age at diagnosis is 1 year). These are associated with a germline mutation of the *RBI* gene.^{2,4} Remaining 60% are sporadic, usually unilateral, and present relatively late (median age at diagnosis is 2 years). These tumours have somatic *RBI* gene mutations. *RBI* is a tumour suppressor gene located on the chromosome 13q14.^{2,4,5}

Most common presenting symptoms are leukocoria, strabismus, and proptosis. When the tumour is exophytic, it may lead to non-rhegmatogenous retinal detachment and consequently sudden painless loss of vision in the affected eye. Intracranial extension is relatively rare but when present can result in hydrocephalus leading to headaches and increased susceptibility to seizures.^{2,6} Our patient presented with headache (due to hydrocephalus) and loss of vision in both eyes (due to involvement of optic chiasma).

Imaging is imperative for pre-treatment evaluation. Ultrasound confirms the diagnosis by demonstrating an irregular intraocular mass which is usually more echogenic than the vitreous body and contains foci of calcifications, which are easily recognised by the posterior acoustic shadows. Cross-sectional imaging such as CT and MRI helps to delineate the true extent of the tumour as well as identify if there is a coexistent retinoblastoma in the apparently normal eye (since small retinoblastomas can be missed by clinical examination alone) and whether a pineoblastoma is present in addition to the retinoblastoma (so-called trilateral retinoblastoma).³

MRI, especially contrast enhanced MRI, is superior to other imaging modalities to evaluate the extent of optic

nerve involvement, extraocular & intracranial extension, and leptomeningeal spread. Vitreal and subretinal seeding may also be detected by high resolution fast spin echo sequences although their absence at MRI does not exclude their presence.^{7,8,9} In our case, the tumour had spread via the optic nerve to involve the optic chiasma. There were no obvious vitreal or subretinal seeds.

Chemoradiotherapy was offered as the primary treatment modality but the parents declined and the patient was subsequently lost to follow up.

Conclusion

Imaging, especially contrast enhanced MRI, is invaluable for pre-operative staging of the disease and evaluating the prognosis & potential for vision in the affected eye. So the current recommendation for any newly diagnosed case of retinoblastoma is to undergo an MRI for early detection and proper evaluation. Treatment of retinoblastoma is multidisciplinary and multimodal, and includes surgery, radiotherapy, cryotherapy, and chemotherapy, depending on the clinical context. A tumour confined within the eyeball has a survival rate of 90% at 5 years while mortality reaches over 90% if tumour extended outside the eyeball.^{9,10}

Conflict of Interest: None

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