Retinoblastoma with significant intravitreal haemorrhage: a rare presentation

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DESCRIPTION

Retinoblastoma is the most common intraocular tumour of the eye in children. It constitutes 11% of all cancers within the first year of life. In one-third of cases, it occurs bilaterally; usually, before 5 years of age without any gender predilection.^{1,2} Trilateral retinoblastoma term is used for unilateral or bilateral presence associated with intracranial tumours of the pineal gland.^{3–5} Retinoblastoma can be hereditary or sporadic—two-thirds of patients present with sporadic mutation of the RB1 gene; located on the long arm of chromosome 13 (13q14). Retinoblastoma is classified as primitive neuroepithelial neoplasm, which arises from the neural epithelium that is typically programmed to evolve into retinal photoreceptor cells.⁶ The primary sign of retinoblastoma is a white pupillary reflex (leukokoria) which is a white or cloudy and dilated pupil. The pupil may look white at certain angles or red in a photo that uses a flashlight, however mostly, the pupils are of similar sizes. Other presentations include crossed eyes (strabismus), eyes may appear misaligned, ranging from mild to severe. Inflammation and irritation can occur anywhere in the eye resulting in redness of the eye. The child can also present with decreased vision (bilateral), retinal detachment, glaucoma, hypopyon or ocular pain.

The primary imaging investigations for retinoblastoma are CT and MRI. CT helps us identify intralesional calcification. MRI is more valuable in retinoblastoma to detect the extension via the optic nerve, leptomeningeal involvement and metastasis in the brain parenchyma.⁷ The tumour may disseminate in the subarachnoid space via the optic nerve.

The mainstay treatment for retinoblastoma is enucleation, but nowadays, conservative management such as cryoablation, photocoagulation and chemotherapy are also recognized for smaller lesions.

Intraocular retinoblastoma has a better prognosis than extraocular or metastatic retinoblastoma. Rarely, spontaneous regression with resultant cure and shrinkage of the eyeball may occur due to necrosis followed by calcification. Prognosis is fair (survival rate 70%–85%) if the eyeball is enucleated before the occurrence of extraocular extension.

A 2-year-old girl came to the outpatient department with a 1-week history of left eye pain, redness, vision loss and headache. Her physical examination revealed mild erythema around the left periorbital tissues. The patient had no history of any ocular injury, significant medical history or positive family history. Ophthalmic examination revealed an impaired visual acuity of only hand motion detection in the left eye. Slit-lamp examination

Figure 1 MRI orbit. (A) Unenhanced T1W image in axial plane demonstrates increased signals in the posterior chamber of left eye globe compared with normal hypointense signals in the right eye globe (white arrow). (B) T2W image in axial plane shows fluid-fluid levelling due to intravitreal haemorrhage (black arrow) in the posterior chamber of left eye globe with internal heterointense lesion attached to the posterior part of the globe (white arrow). (C) Susceptibility-weighted image in axial plane depicts blooming artefact in the lesion suggestive of calcification (white arrow). (D, E) Contrast-enhanced T1W images in axial and coronal plane demonstrate heterogeneously enhancing lesion in the posterior chamber of the left eye globe, reaching up to the posterior surface of the lens (white arrow). (F) Diffusion-weighted MRI in the axial plane demonstrates diffusion restriction in the lesion which suggesting hypercellularity of the lesion (white arrow).

demonstrated no abnormalities in the anterior segment. Fundus examination displayed vitreous haemorrhage, candle-wax spots and enlarged tortuous vessels. B-scan ultrasonography showed irregular ill-defined hypoechoic lesions located in the posterior chamber with multiple low-level echoes. Intraocular pressure was 14 mm Hg in both eyes. Visual acuity and examination of the right eye

Patient's perspective

I was distressed to see my daughter in pain and sudden vision loss and seeing her in such a life-threatening situation. We came to the hospital with great hopes of saving her life. Doctors assured us about the condition of the patients and made us understand that the left eye is in grave condition and we are supposed to remove it to save the baby's life. After lots of discussions, we agreed to remove the left eye. Though it is quite traumatic to see her with a single eye, we are happy doctors could save my daughter's life. Doctors also asked us to come for a general check-up every month.

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Learning points

- ▶ The posterior chamber lesion may be masked under the direct ophthalmoscopic examination due to a significant amount of vitreal haemorrhage.
- ▶ Radiological examination plays an imperative role in the diagnosis of retinoblastoma as it guides us on the lesion characteristic and extension.
- ▶ MRI orbit is the ideal investigation for retinoblastoma because of its inherent high soft-tissue resolution and to rule out the extension of the disease in the subarachnoid space via the optic nerve.

were normal. Routine investigation of bone marrow, cerebrospinal fluid and blood was unremarkable.

MRI scan with contrast of the brain and orbit was performed for further evaluation, which elicited abnormal high-intensity signals in the left eye globe on T1 images, differential fluid signal intensities forming fluid-fluid, levelling on T2-weighted images, along with a heterointense lesion in the posterior chamber. Susceptibility-weighted images showed blooming in the lesion suggestive of calcification. Diffusion-weighted images demonstrate restricted diffusion suggesting the hypercellularity of the lesion. The postcontrast study revealed an enhancing, irregular intraocular mass within the posterior chamber of the left globe. The optic nerve head did not demonstrate abnormal enhancement. No definitive evidence of the left optic nerve

involvement or intracranial extension was seen on the MRI scan (Figure 1A–F). The right eye globe was normal.

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REFERENCES

- 1 Amoaku WM, Willshaw HE, Parkes SE, et al. Trilateral retinoblastoma. A report of five patients. *Cancer* 1996;78:858–63.
- 2 Young JL, Smith MA, Roffers SD. Retinoblastoma. In: Reis LAG, Smith MA, Gurney JG, eds. *Cancer incidence and survival among children and adolescents: United States SEER program 1975-1995*. Bethesda, MD: National Institutes of Health, 1999.
- 3 Blach LE, McCormick B, Abramson DH, et al. Trilateral retinoblastoma--incidence and outcome: a decade of experience. *Int J Radiat Oncol Biol Phys* 1994;29:729–33.
- 4 De Potter P, Shields CL, Shields JA. Clinical variations of trilateral retinoblastoma: a report of 13 cases. *J Pediatr Ophthalmol Strabismus* 1994;31:26–31.
- 5 Provenzale JM, Gururangan S, Klintworth G. Trilateral retinoblastoma: clinical and radiologic progression. *AJR Am J Roentgenol* 2004;183:505–11.
- 6 Towbin RB, Bisset GS. Pediatric case of the day. trilateral retinoblastoma. *Radiographics* 1986;6:1100–4.
- 7 Smirniotopoulos JG, Bargallo N, Mafee MF. Differential diagnosis of leukokoria: radiologic-pathologic correlation. *Radiographics* 1994;14:1059–79.

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