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Primary intraocular malignant rhabdoid tumor presenting as orbital mass with intracranial extension in an adolescent

A malignant rhabdoid tumor (MRT) is a highly aggressive tumour of the kidney. Extra renal MRTs have also been described as cytologically resembling renal rhabdoid tumours and are known to be highly lethal with a poor prognosis. Orbital presentation of MRT was first reported by Rootman et al. in 1989.[1] Since then, a few MRTs of the orbit have been reported in infants, but none as a primary intraocular MRT presenting as an orbital mass in adolescents.

One such case has been reported with clinicoradiological features, histopathology, and immunohistochemical characteristics. The case describes a 13-year-old male patient with a rapidly progressive proptosis and a mass protruding from his left eye starting 5 months previously. His records showed visual acuity of no light perception OD and a hyperechoic intraocular lesion on ultrasound B-scan. At presentation, a fungating mass which was firm and congested and had an irregular surface and margins was identified in his left eye (Fig. 1A). The patient's right eye was normal with 20/20 vision. Contrast-enhanced magnetic resonance imaging (MRI) of the orbits showed a heterogeneous enhancing mass filling the eyeball and extending into the orbit. Contiguous involvement of the optic nerve extending to the optic chiasma was also noted (Fig. 2A, 2B). Visual field tests revealed a superotemporal quadrantanopia in the patient's right eye, but systemic examination did not show any abnormality. Other tests, such as blood biochemistry, ultrasound of the abdomen and chest x-ray, were normal.

An incision biopsy was taken from the orbital mass and revealed a poorly differentiated malignant tumour consisting of cells with abundant cytoplasm and pleomorphic nuclei and nucleoli (Fig. 3A). Immunohistochemical stains were positive for cytokeratin and vimentin and negative for desmin, leucocyte common antigen, synaptophysin, neuron specific enolase, and HMB-45 (Fig. 3B). These findings were suggestive of a malignant rhabdoid tumour.

The patient received 3 cycles of chemotherapy consisting of ifosfamide and etoposide, initially resulting in a significant reduction of the tumour mass (Fig. 1B). Exenteration was offered after tumour reduction, but the patient refused surgery. Tumour progression and central nervous system (CNS) invasion along the optic nerve was consequently noted, and the patient succumbed to the tumour.

Primary intraocular presentation of MRT is extremely rare. Akhtar et al. reported a case in which MRT of the

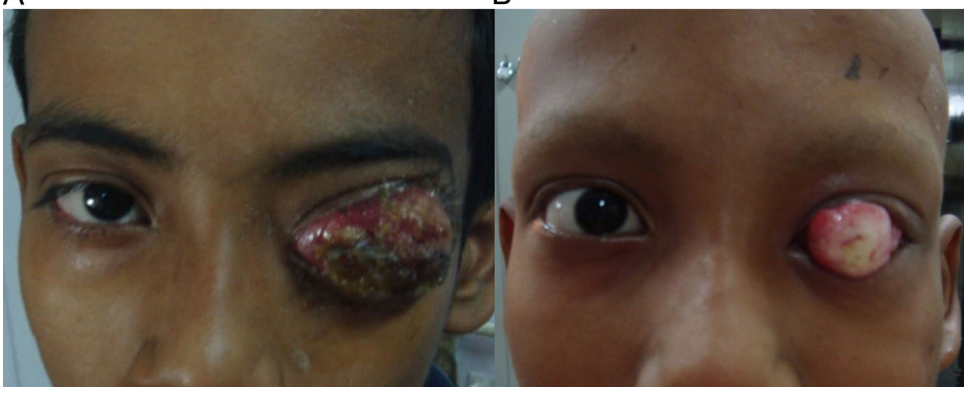

Fig. 1—A, Clinical photograph showing a fungating mass protruding from the left eye. B, Clinical photograph showing reduction in the size of the mass in the left eye following chemotherapy.

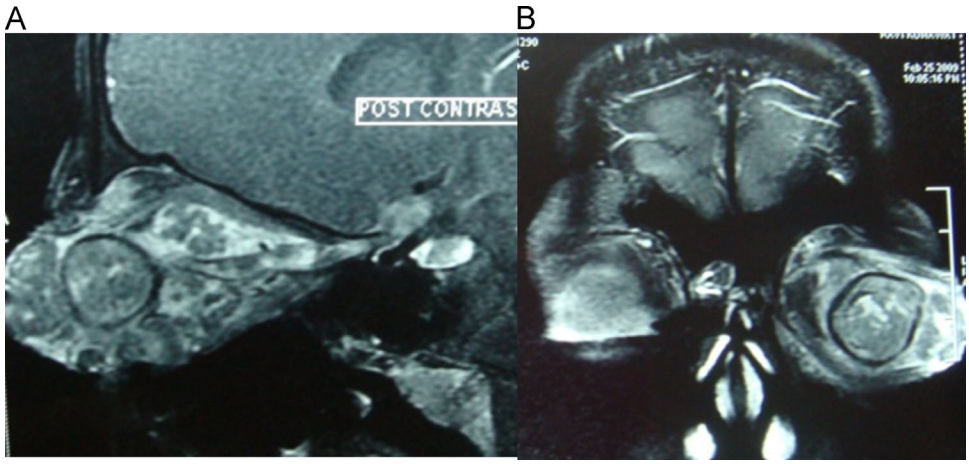

Fig. 2—A, Sagittal section of post-contrast magnetic resonance imaging (MRI) scan showing a heterogeneous enhancing mass occupying the entire left orbit with involvement of the optic nerve up to the optic chiasma. B, MRI, coronal section showing a staphylomatous globe filled with an intraocular mass with extrascleral extension in the orbit.

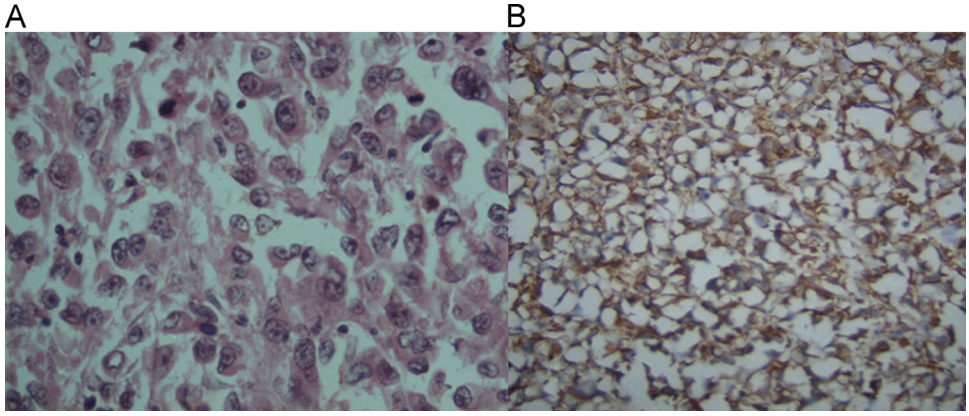

Fig. 3—A, Microphotograph showing large round to oval tumour cells with marked pleomorphism and prominent nucleoli (hematoxylin and eosin stain, 400×). B, Immunohistochemical stain vimentin showing strong positivity in the tumour cells (400×).

kidney was diagnosed as an intraocular metastasis in a 2-week-old neonate.[2] The patient's eye was enucleated on the presumptive diagnosis of retinoblastoma. Another MRT case presenting as an intraocular mass was reported in a 1-month-old infant by Shah et al., possibly resulting from a metastasis of an undetected primary tumour.[3] In the present case, MRT presented as a fungating mass primarily arising from the eye, with no evidence of a primary tumour elsewhere in the body. Our case presented in a patient aged 13 years, a much older age compared to the reported age of intraocular cases (mean age 3 ± 1.4 weeks) and orbital cases (mean age 10.37 ± 14.5 months) of MRT in children.[1–10] In this scenario, the clinical picture resembles other tumours of childhood and adolescence such as a retinoblastoma, rhabdomyosarcoma, or medulloepithelioma. The diagnosis is established based on histopathology and immunohistochemistry.

The term rhabdoid implies a resemblance to rhabdomyoblasts, which is characteristic of aggressive, dedifferentiated malignancies. On microscopic examination, rhabdoid tumours consist of polygonal cells with eosinophilic globules, displacing notched or vesicular nuclei that have prominent nucleoli. Immunohistochemistry shows vimentin, a wide spectrum of keratin, and epithelial membrane antigen as the most consistent antigenic phenotypes. In our case, the diagnosis of MRT was based on the pathological finding of immunohistochemical co-expression of cytokeratin and vimentin in an undifferentiated tumour. Although MRT was the most probable diagnosis in this patient, it is important to note that such co-expression can also manifest in other tumors, such as epithelioid sarcoma, angiosarcoma, metastatic renal and thyroid carcinoma, and biphasic mesothelioma.

Extrarenal MRTs in the orbit are typically resistant to multimodal therapy.[5–7] Recurrences and systemic metastasis have been reported following exenteration combined with chemotherapy, despite the lesion being confined to the orbit.[5,6] Watanabe et al. showed promising results with gamma-knife radiosurgery after high-dose chemotherapy.[9]

Ifosfamide, in combination with carboplatin and etoposide, was found to be effective in reducing the tumour burden prior to surgical resection.[7,9] However, the role of radiotherapy is not established. In our case, chemotherapy with ifosfamide and etoposide showed remarkable tumour reduction initially but early CNS invasion along the optic nerve lead to the patient's death in our case.

To the best of our knowledge, this case represents the first report of primary intraocular MRT presenting as an orbital mass in adolescence. We therefore emphasize that malignant rhabdoid tumours, although rare, should be considered in the setting of a rapidly progressive intraocular mass with orbital extension in this age group.

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Cat-scratch penetrating globe injury with inadvertent fistula and hypotony

To our knowledge, this is the first case in the literature of a cat-scratch globe injury causing an inadvertent delayed-onset filtering bleb with hypotony and decreased vision that necessitated repair with a corneal patch graft. Untreated hypotony can result in maculopathy, cataract, corneal edema, and choroidal detachment. There are currently 4 published case reports on cat-scratch corneal lacerations in the United States, but none featured an inadvertent fistula or delayed hypotony.

A 32-year-old Caucasian female with no ocular history presented to the University Hospital complaining of blurry vision and pain in her right eye after her cat scratched her eye while she was sleeping. During surgical repair, a 2-mm full thickness limbal laceration was noted at 3 o'clock with iris prolapse through the wound. One week postsurgery, the patient's visual acuity remained blurred at 20/200, and intraocular pressure was 4 mm Hg.

A small cyclodialysis cleft was then noted at 3 o'clock with gonioscopy. During 7 months of regular follow-up, intraocular pressure remained persistently low (3–8 mm Hg), reaching a nadir of 2 mm Hg approximately 2 months postinjury. Visual acuity gradually improved to 20/40 at 2 months postsurgery and 20/20 thereafter. Additional consultation revealed a Seidel negative inadvertent nasal filtering bleb with a clear communication apparent on gonioscopy of the nasal angle. The bleb subsequently became Seidel positive, necessitating urgent repair.

During the surgical repair, a 2-mm scleral defect was noted at the site of the prior globe rupture in the superior nasal quadrant, through which aqueous was freely percolating. To approximate the scleral defect, 10-0 nylon sutures were placed (Figs. 1 and 2). After suture placement, a gap remained at the site of injury and a corneal patch graft was fashioned to cover the defect. A trapezoidal shape graft was fashioned with the anterior edge and curved to match the limbal contour (Fig. 3). The corneal graft was split approximately half thickness with the removal of endothelial and epithelial layers. The half thickness stromal tissue was used to cover the defect and was secured in place with 10-0 nylon sutures. Four weeks postprocedure, the patient's intraocular pressure improved to within normal limits, and she remained asymptomatic for the following 3 years with best corrected visual acuity of the right eye at 20/15.

Estimates of animal-related ocular injuries per year in the United States vary widely.[1] Of domestic animals, dog bites and cat scratches account for most injuries. In the context of periorbital and globe wounds, the mechanisms of injury differ. While dog bites cause gross tissue damage and bone fractures, cat claws cause more localized wounds, often tearing through full thickness cornea.[2,3] Eighty percent of cat scratches and bites are from an animal known or belonging to the victim, and the most common