Abstract:
Introduction—Retinoblastoma is rare after 3 years of age, the mean age of diagnosis being 18 months in bilateral retinoblastoma and 24 months in unilateral cases. We report a 13 year old girl with unilateral retinoblastoma and intracranial extension. Case History—The girl presented with loss of vision in the left eye and prominence of the left eye along with pain for 2 months duration. Clinical examination of the left eye revealed no perception of light, proptosis, shallow anterior chamber, neovascular glaucoma and total cataract. A review of imaging done 10 months prior to presentation showed a heterogeneous intraocular mass partially filling the globe with no optic nerve involvement. A more recent imaging showed the tumour completely filling the globe with optic nerve involvement up to the chiasm. The differential diagnoses included optic nerve glioma and meningioma with intraocular involvement, retinoblastoma with intracranial extension and an intraocular inflammation with extension. The eye was enucleated and histopathological examination showed malignant round cell tumour consistent with retinoblastoma. Conclusion—The differential diagnoses and relevant literature review of an intraocular tumour with wide spread involvement is discussed. Although rare, retinoblastoma should be considered as a differential diagnosis for an intraocular tumour with wide spread involvement in a teenager.

Keyword: Retinoblastoma, teenager

INTRODUCTION
Retinoblastoma is a rare disease, the incidence being approximately 1:15,000 to 1:20,000 live births (1). It is rare to find patients with retinoblastoma after the period of early childhood, as 90% are diagnosed before the age of 5 years (2). We report a case of advanced retinoblastoma with intracranial extension in a teenage girl, who had an atypical presentation of the disease. CLINICAL FEATURES A 13 year old girl presented to us with history of headache of 2 months duration. She also complained of pain and redness of her left eye for the same duration. She had noticed a white reflex in the left eye 10 months prior to the present complaints, for which she had been seen and investigated elsewhere. She gave history of decreased vision in the left eye over the past 10 months, with a complete loss of vision in the left eye for the past 2 months. A general systemic examination revealed enlarged left pre-auricular and sub-mandibular lymph nodes which were non-tender and mobile. There were no peripheral markers of neurofibromatosis. Ocular examination of the right eye revealed a vision of 6/6 (Snellen Visual Acuity Chart) with normal anterior and posterior segments. The left eye had no perception of light. The left eye showed axial proptosis. There was circum-ciliary congestion. A diffuse microcystic epithelial edema was present throughout the cornea. The anterior chamber was shallow; with peripheral irido-corneal touch all around for 360°. The iris showed large, irregularly branching new vessels coursing over the entire surface of the iris. The pupil was dilated and fixed with a total afferent pupillary defect. There was a total cataract, obscuring view to the fundus. The intraocular pressure was 12mmHg in the right eye and 28mmHg in the left (Goldmann Applanation Tonometry).

IMAGING

Figure 1: CT scan showing calcific spots within the left intraocular tumor
need for histopathological diagnosis before initiating further treatment.

enucleate the left eye as it was a painful blind eye and there was a Retinoblastoma Group E was made. A decision was taken to examination and the 2 sets of imaging, a clinical diagnosis of (Figure 2). HISTOPATHOLOGY On considering the history, involved with the tumour, which extended upto the optic chiasm showed the same mass to be filling the globe. The optic nerve was same time showed that the mass was hypointense on T2-weighted images and partially filling the globe. A more recent imaging (MRI) the same mass to be filling the globe. The optic nerve was involved with the tumour, which extended upto the optic chiasm (Figure 2). HISTOPATHOLOGY On considering the history, examination and the 2 sets of imaging, a clinical diagnosis of Retinoblastoma Group E was made. A decision was taken to enucleate the left eye as it was a painful blind eye and there was a need for histopathological diagnosis before initiating further treatment.

Figure 2: MRI showing tumor filling the globe & involving the entire optic nerve
An ocular ultrasonography revealed the vitreous cavity of the left eye to be filled with dot-like echoes, suggestive of calcification, as there was presence of shadowing beyond the hyper-echoic spots. A review of imaging done 10 months prior to presentation showed an intraocular mass in the posterior segment of the left globe. This mass had calcific spots, which was seen in the plain CT scan (Figure1). The mass enhanced on contrast. The optic nerve was normal. An MRI done at the same time showed that the mass was hypointense on T2 weighted images and partially filling the globe. A more recent imaging (MRI) showed the same mass to be filling the globe. The optic nerve was involved with the tumour, which extended upto the optic chiasm (Figure 2). HISTOPATHOLOGY On considering the history, examination and the 2 sets of imaging, a clinical diagnosis of Retinoblastoma Group E was made. A decision was taken to enucleate the left eye as it was a painful blind eye and there was a need for histopathological diagnosis before initiating further treatment.

Figure 3: HPE showing necrotic tumor filling the globe & involving the choroid
Histopathological examination revealed a large necrotic tumour in the posterior segment of the enucleated globe pushing the lens forward. There was involvement of the choroid and the optic nerve with tumour (Figure 3). The tumour was composed of closely packed small cells with round, hyperchromatic, mitotically and apoptotically active nuclei with scanty cytoplasm (Figure 4). Immuno histochemistry revealed that these cells were immuno positive for Neuron-specific enolase and synaptophysin, but negative for other round-cell tumour markers (GFAP, CD3 and CD7a). Hence, a histopathological diagnosis of retinoblastoma was made. FURTHER COURSE On follow-up, the patient was pain-free after enucleation. Cosmetic rehabilitation was done by the fitting of an ocular prosthesis. All her family members were screened by dilated fundus examination and found to be negative for retinoblastoma. The patient was evaluated by Paediatric Oncology and started on palliative chemotherapy (VEC - vincristine, etoposide and carboplatin).

DISCUSSION:
The various causes of proptosis in a teenage patient include inflammation, developmental cysts, haematoma and tumours. Among the various possibilities, the differential diagnoses considered for our patient were optic nerve glioma or meningioma with ocular involvement, retinoblastoma with intracranial extension or an inflammatory orbital mass with intraocular and intracranial extension. Optic nerve glioma was considered initially as a likely diagnosis for our patient because this tumour is common among young girls. Moreover, the spread of the tumour along the optic nerve upto the chiasm is characteristically seen in optic gliomas. Typically, patients with optic nerve glioma present with a gradual, painless visual loss. Optic gliomas have been reported to have intraocular involvement (3) (4) (5). However, in none of these cases has the intra-ocular part of the tumour filled the entire posterior segment of the globe. Furthermore, in our patient, the imaging showed a mass which was isointense on T1 and hypointense on T2 weighted images. Optic gliomas are typically hypointense on T1 and hyperintense in T2 images. The presence of calcific spots in the mass pointed against a glioma, as calcification is rare in optic gliomas (6). Optic nerve meningiomas have also been reported to have intraocular involvement (3) (4) (5). However, in none of these cases has the intra-ocular part of the tumour filled the entire posterior segment of the globe. Furthermore, in our patient, the imaging showed a mass which was isointense on T1 and hypointense on T2 weighted images. Optic gliomas are typically hypointense on T1 and hyperintense in T2 images. The presence of calcific spots in the mass pointed against a glioma, as calcification is rare in optic gliomas (6). Optic nerve meningiomas have also been reported to have intraocular involvement, hence was considered a possibility in our patient (7) (8) (9) (10). Meningiomas can present with gradually increasing proptosis. 30% of them are associated with the presence of optociliary shunt vessels. CT scan shows the presence of thickening of the optic nerve and the presence of calcification. They are hyperintense in T1 weighted Images and hypointense in T2 weighted images, which is in fact similar to the images of our case. However, these tumours are commonly seen in middle-aged women, so was a less likely diagnosis in our teenage patient. Retinoblastoma is a rare entity. However, it is the most common intraocular malignancy in early childhood (11) (12). The average age of diagnosis of retinoblastoma is 24 months in unilateral cases and 13 months in bilateral cases (1). It is rare to find retinoblastoma presenting in an older child, especially if older than 5 years of age. There have been case reports of retinoblastoma presenting in older children as well as adults (13) (14) (15), the first of these being a report by Verhoeff in 1929 of retinoblastoma in a 48 year old man (16). A case series of retinoblastoma in older children was published, highlighting the atypical presentations seen in these cases.
However, none of the patients were described to have extensive retinoblastoma as seen in a teenager.

Reference:


12 Kanski. Clinical Ophthalmology. 6th Ed.


