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Retinoblastoma Presenting As Coat's Disease

KEYWORDS: Retinoblastoma; Coat's disease; Orbital neoplasm.

Dr. Sujata Tripathi
M.D.Pathology, B-8 Hyderabad estate, Nepeansea road. Mumbai- 36

Dr. Bhagyashree Madavi
M.S. Ophthalmology, Department of Ophthalmology, M.P.Shah medical college, Jamnagar – 361008

ABSTRACT *Coats' disease and retinoblastoma are the two common types of orbital pathology in young children. These two conditions are prognostically different but have overlapping imaging features. We present a case of retinoblastoma to illustrate the specific imaging and histopathologic features, which can help the readers differentiate it from coat's disease, a close differential diagnosis.*

INTRODUCTION

Retinoblastoma (22-24%) and Coats' disease (4-16%) are two common congenital intraocular lesions in children presenting with leukocoria. Both conditions can be associated with calcifications and therefore difficult to differentiate on imaging. The other group consists of persistent hyperplastic primary vitreous (19-28%), retrolental fibroplasias (5-13%), posterior cataract (13.5%), coloboma, congenital posterior choroidal defect (11.5%), larval endophthalmitis (6.5-16%). Abnormalities in this latter group can be diagnosed by imaging with higher confidence.

Retinoblastoma is the most common intraocular tumour in children. It arises from the retina in infancy, and manifests before the age of 5 years. Retinoblastoma accounts for 11% of all cancers in the first year of life. Bilateral or multifocal tumours occur in patients with hereditary retinoblastomas and account for 20 to 34% of all cases. The proportion of bilateral tumours in patients less than a year old is higher. There are hereditary and non-hereditary forms. There is an association with a second malignancy, of which osteosarcoma is the most common. Bilateral inherited retinoblastoma is associated with pineal or parasellar tumours, and is also known as trilateral retinoblastoma.

Coats' disease is a vascular malformation of the retina that produces lipoproteinaceous subretinal exudates. The formation of retinal telangiectasia leads to the breakdown of the retinal-blood barrier at the level of the endothelial cell, allowing leakage of blood products into the retina and subretinal space. This fluid contains cholesterol crystals and lipid-laden macrophages. Over time, the accumulation of this lipoproteinaceous fluid leads to thickening of the retina and causes massive, exudative retinal detachment. The peak prevalence of this condition is at 6 to 8 years of age but the range of presenting ages can be quite wide (5 months to 71 years). There is a male predominance (69-85%), and the disease is usually unilateral (83-95%). If bilateral, one eye is usually minimally affected. By far the most common presenting sign is leukocoria, but patients may also present with strabismus, painful glaucoma, or loss of vision. This condition affects a slightly older age-group than does retinoblastoma.

CASE REPORT

A healthy 4-year-old boy presented with leukocoria of the right eye. On examination, the patient demonstrated poor visual fixation in the right eye and steady fixation in his left eye. During examination under anaesthesia, intraocular pressures were within normal ranges. Ultrasonography confirmed a detached and diffusely thickened retina with no intraretinal calcification. Indirect Ophthalmoscopy revealed an unusual exudative retinal detachment. The retina was diffusely thickened and associated with intraretinal and preretinal haemorrhages, a white flocculent material in between areas of retinal folds, and an anomalous retinal vasculature pattern throughout. Intraoperative fluorescein angiography confirmed this unusual vascular pattern with arterio-venous communication, prominent telangiectatic vessels, leakage, and 3601 of peripheral capillary non-perfusion. Anterior segment evaluation showed diffuse iris hyperfluorescence consistent with iris neovascularization. Brain magnetic resonance imaging (MRI) was performed and showed partial retinal detachment with focal thickening at the lateral aspect which was T1 iso-to hyper-intense and T2 hypointense in signal.

The differential diagnosis included Coats' disease, persistent hyperplastic primary vitreous, retinal dysplasia, and an atypical presentation of retinoblastoma. Given the uncertainty over the clinical diagnosis and the poor visual prognosis, enucleation of the eye was performed. Pathologic study revealed an endophytic mass arising from the retina extending from ora to ora, with vitreous seeding and extensive exudate in the subretinal space. Cells exhibiting a unique form of differentiation to produce elements similar to those seen in photoreceptor cells were seen. Three cytologic features of rosette formation were seen: fleurettes, Homer-Wright rosettes, and Flexner-Wintersteiner rosettes. Flexner-Wintersteiner rosettes, or true rosettes, are nearly pathognomonic for retinoblastoma. They are characterized by differentiated tumor cells arranged around a patent central space (figure 1).The diagnosis of retinoblastoma with Coat-like response was confirmed

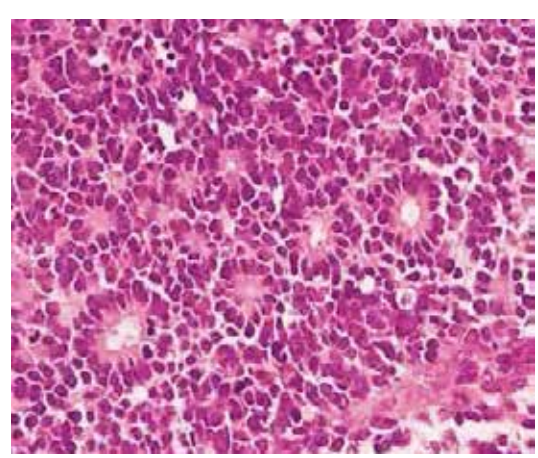

Figure 1 : Flexner wintersteiner rosettes – pathognomic of retinoblastoma on histopathological examination

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DISCUSSION

Retinoblastoma and Coats' disease, though prognostically different, can have overlapping clinical and imaging features, which makes diagnosis difficult. However, making a correct diagnosis is crucial to providing the necessary treatment. For retinoblastoma, it is important to avoid any risky prior intraocular procedure before the definitive enucleation, as this might accelerate tumour seeding. From previous studies, as many as 58% of the patients with Coats' disease underwent unnecessary enucleation, and as many as 16% with a retinoblastoma were misdiagnosed as Coats' disease.

Overall, there remains an overlap of clinical and imaging features between these 2 entities. By and large, Coats' disease has T1 and T2 hyperintensity, linear enhancement post-contrast, a smaller globe volume, and a lipid peak on MR spectroscopy. Calcification is an uncommon radiographic features in Coats' disease, it may be present when the disease is advanced. On the contrary, the majority of retinoblastomas are T1 hyperintense and T2 hypointense, exhibit masslike enhancement with a normal and symmetrical globe volume compared to the contralateral eye. In retinoblastoma, calcification is very common — thick and coarse. The following features provide some clues, which might help arrive at the correct diagnosis

CONCLUSION

Retinoblastoma is a highly malignant tumor of the eye. Early diagnosis and prompt treatment are important factors in a child's survival and can often salvage useful vision in one or both eyes,atypical manifestations of retinoblastoma such as hyphaema, neovascularization and pre retinal haemorrhages can pose diagnostic challenges therefore investigative modalities such as ultrasonography, MRI and histopathology are all important and must be performed at the earliest to arrive at a conclusive diagnosis.

It is good practice to maintain a high level of suspicion for retinoblastoma in all children presenting with unusual presentation of ocular diseases.

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