Adult Onset Retinoblastoma: A Case Report

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Introduction

Retinoblastoma usually presents in children younger than 5 years. Adult retinoblastoma is rare, and diagnosis is usually made at a fairly advanced stage. We report a case with whitish, elevated, vascularized retinal mass and vitreous seeding diagnosed as undifferentiated retinoblastoma.

Case Report

A 45-year-old male presented to Peking Union Medical College Hospital (PUMCH) on January 23, 2013. He began to notice floaters in the left eye 1 year before, and was diagnosed with "glaucoma OS" in local hospital. Intraocular pressure (IOP) decreased from 36 mmHg to normal with topical IOP-lowering medication. The patient's visual acuity of the left eye began to decrease in October, 2012. He was diagnosed with "neovascular glaucoma OS" in local hospital, and was advised to have operation. During pre-operative examination, an intraocular mass was found.

On presentation to PUMCH, best corrected visual acuity was 1.2/Jr4 OD and 0.5/Jr7 OS. IOP was 18 mmHg OD and 42 mmHg OS. In the left eye, there were anterior chamber cells, neovascular of the pupil margin, and plenty of small white floaters in the vitreous [Figure 1a]. Fundus examination showed large, whitish, elevated mass lesions with vascular in the inferonasal quadrant of the retina [Figure 1b] and on the disk, with retinal seedings of whitish, nodular lesions [Figure 1c]. B-scan ultrasonography showed vitreous opacities and intraocular elevated lesions with moderate reflective echoes and absence of calcification [Figure 1d]. Systemic examinations including blood and urine tests, electrocardiogram, chest computed tomography, abdomen B-scan ultrasonography, as well as 99mTc-methylene diphosphonate whole body bone imaging were normal.

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After topical IOP-lowering medication and intravenous mannitol treatment, IOP OS was out of control. A clinical diagnosis of suspected retinoblastoma was made. As adult cases were rare, diagnostic vitrectomy and retinal lesion biopsy were performed in the left eye on January 29, 2013. Vitreous cytology showed small malignant cells. Histopathological examination of retinal lesion showed small cell neuro endocrine carcinoma. Immunohistochemistry staining: Syn (+), CD56 (NK-1) (+), AE1/AE3 (−), CgA (−), S-100 (−), Desmin (−), Ki-67 (index 8%). As malignant retinal tumor with vitreous seeding of the left eye was diagnosed, the patient was informed sufficiently and decided to receive enucleation. Left globe enucleation and hydroxyapatite orbital implant implantation was performed on Febuary 7, 2013. Histopathological examination showed small cell neuro endocrine carcinoma of the left eye [Figure 2]. Optic nerve and sclera were not involved. Immunohistochemistry staining: CgA (+), Syn (+), NSE (+), CD56 (NK-1) (+), S-100 (−), GFAP (−), HMB45 (−), LCA (−), Melan-A (−), p53 (−), Ki-67 (index 20%). The final diagnosis was undifferentiated retinoblastoma OS.

Discussion

Retinoblastoma may be present at birth is usually diagnosed between birth and 5 years of age. The tumor may involve one or both eyes, initially as one or more white masses usually in the posterior pole of the eye.[1] When an adult patient presents with intraocular whitish masses, retinoblastoma usually will not be considered first, because of its relatively low frequency. Differential diagnosis includes metastatic carcinoma, amelanotic melanoma, astrocytoma, lymphoma, endophthalmitis, and retinal inflammatory diseases. Intraocular medulloepithelioma is another lesion that should be differentiated from retinoblastoma. It is a rare congenital tumor derived from the nonpigmented ciliary epithelium, and

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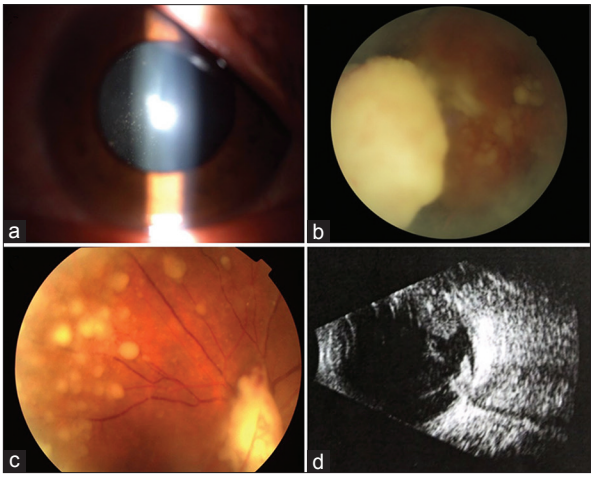

Figure 1: Slit-lamp examination showed plenty of small white floaters in the vitreous (a). Fundus examination showed large, whitish, elevated mass lesion in the inferonasal quadrant of the retina (b). Fundus examination showed whitish, elevated mass lesion with vascular on the disk, with retinal seedings of whitish, nodular lesions (c) B-scan ultrasonography showed vitreous opacities and intraocular elevated lesions and absence of calcification (d).

usually occur between 5 and 20 years of age. Final diagnosis depends on histopathology examination.

On literature review from Pubmed, we found about 30 cases of adult retinoblastoma, aging from 20 to 74 years old.[2-4] Almost all cases were sporadic and unilateral. Clinical presentation included diminution or loss of vision, flashes, floaters, and photopsia. Leukocoria is a typical manifestation in children's retinoblastoma, but not a common presentation in adults. Characteristic manifestation of these adult retinoblastoma was whitish, elevated, vascularized retinal mass with vitreous seeding. In a few cases, the masses involved the whole eye. Diffuse retinoblastoma was also found.[5] Calcification is a characteristic of retinoblastoma in children, not an important finding in adults, with only a few patients showing calcification in imaging studies. In the majority of cases, diagnosis were made at a fairly advanced stage. Enucleation was the primary modality of treatment. A few patients underwent external beam radiotherapy, but the results were poor because of the advanced stage of the disease at diagnosis.

Histopathology of the enucleated specimen confirms the diagnosis and identifies the level of differentiation. The majority of the reported retinoblastoma in adults

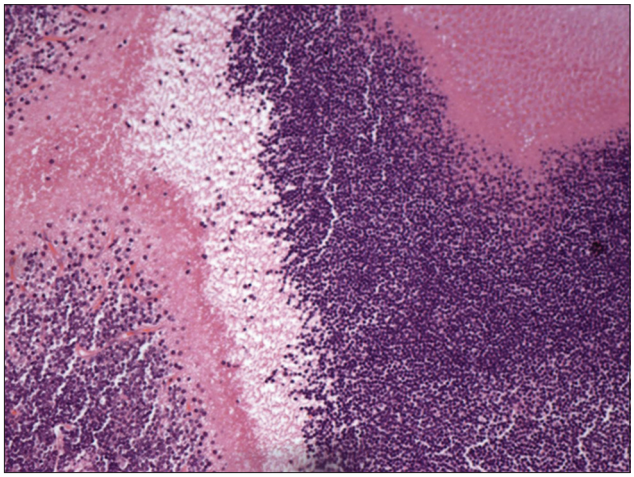

Figure 2: Photomicrograph of the enucleated globe showing small undifferentiated cells with hyperchromatic nuclei and scanty cytoplasm (Hematoxylin and Eosin, ×200).

were differentiated showing Flexner-Wintersteiner rosettes. Our patient's tumor was classified as undifferentiated retinoblastoma with no rosettes and fleurettes, so immunohistochemistry was needed for diagnosis.

Retinoblastoma is rare in adult, but in the presence of whitish mass lesions in the fundus, clinicians should consider it as a possible diagnosis.

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