

A ten-year follow-up of retinoblastoma in an adult: first case in Colombia

Acompanhamento de dez anos de retinoblastoma em um adulto: primeiro caso na Colômbia

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ABSTRACT

Retinoblastomas are the most common primary intraocular malignancy of childhood, with a high incidence in the first 2 years of life. This is the report of the first unilateral retinoblastoma case in a 57-year-old Colombian female, who presented to consultation with an advanced neovascular glaucoma in the left eye. Examination revealed an amelanotic intraocular mass that required enucleation. The histopathology report and specific tumor markers of the enucleated specimen revealed the diagnosis. A 10-year follow-up was carried out with magnetic resonance imaging, with no findings of bilaterality or recurrence in the anophthalmic cavity.

RESUMO

Retinoblastoma é a neoplasia intraocular maligna mais comum da retina na infância, com alta incidência nos primeiros dois anos de vida. Descrevemos o primeiro caso na Colômbia, de retinoblastoma unilateral em uma paciente de 57 anos, que veio à consulta com glaucoma neovascular avançado. Ao exame, apresentava uma massa intraocular amelanótica esquerda, e precisou ser enucleada. O diagnóstico foi confirmado por histopatologia e marcadores tumorais específicos. Foi realizado seguimento por 10 anos, com ressonância magnética, sem achados de bilateralidade ou recidiva na cavidade anoftálmica.

INTRODUCTION

Retinoblastomas (OMIM 180200) are malignant retinal tumors frequently affecting children, with an incidence of one per 15,000 to 20,000 live births.⁽¹⁾ This malignancy originates from mutations of the retinoblastoma or RB1 gene, located in the 13q14 chromosomal region, which are highly penetrant and show a dominant autosomal pattern. Ninety percent of retinoblastomas are diagnosed in children under 5 years of age; in adults, this is a rare pathology, and not considered in differential diagnosis of intraocular tumors.⁽²⁾

This manuscript reports the first unilateral retinoblastoma in adult, in Colombia, diagnosed by immunohistochemistry, as per the World Health Organization (WHO) ethical guidelines, with informed consent given and signed by the patient, and approved by the Bioethics Committee of the Universidad de Antioquia.

CASE REPORT

A 57-year-old female patient, with no significant past medical history, referred non-painful visual acuity decrease on her left eye (OS) during the past 12 months, with no other associated ocular symptoms.

On ophthalmologic examination, right eye (OD) visual acuity was 20/25+ and OS counting fingers at 1 m (three feet). Documented retinoscopy was OD: +1.00 -0.50 x167°, OS: +2.00 -1.50 x76°. No pathological signs in the OD, with intraocular pressure of 10 mmHg. Left eye with conjunctival hyperemia, clear cornea, broad anterior chamber, 360° uveal ectropion, diffuse and generalized rubeosis iridis, corticonuclear cataract grade 2 to 3, and intraocular pressure of 38 mmHg. Right eye with no fundus alterations but OS presented diffuse vitreous hemorrhage, bullous retinal detachment from the nasal quadrant up to the optic nerve, and a non-pigmented lesion of the nasal periphery with hard to observe details due to the vitreous opacity.

The patient brought an ocular ultrasound showing an upper nasal dome-shaped lobulated mass measuring 9.5 mm x 12 mm, homogeneous, with medium to low internal reflectivity (26%), internal calcification, and adjacent retinal detachment (Figure 1A to 1B). The initial diagnosis was peripheral intraocular tumor associated to bullous retinal detachment, vitreous hemorrhage, and neovascular glaucoma. Imaging studies to investigate tumor associated to systemic disease, such as brain magnetic resonance imaging (MRI) and chest X-rays, presented no changes.

Because of associated ocular damage and impossibility to treat by other means, OS enucleation with

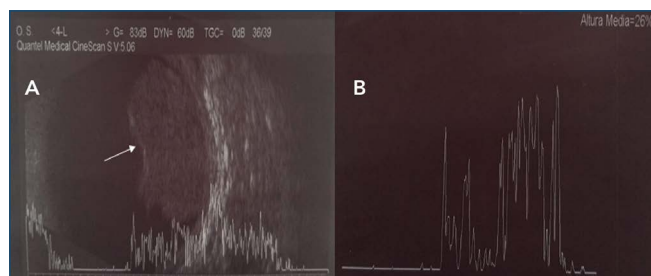


Figure 1. Ocular ultrasound: B mode showing lobulated mass (arrow) with homogeneous internal structure (A). In mode A, medium-to-low internal reflectivity was found (B).

porous polyethylene (Medpor®, Porex Surgical, Newnan, GA, United States) ocular implant was performed. A white, soft, lobulated mass, measuring 12 mm in diameter and 7-mm thick was attached to the retina, and removed for histological examination. The sections were stained by hematoxylin and eosin (H&E) and showed a well-differentiated tumor with Homer-Wright rosettes (Figure 2) with no extension to the sclera, choroids or optic nerve. Immunohistochemistry stains were used to confirm diagnosis. The tissue was strongly positive for neuron-specific enolase (NSE) and positive for synaptophysin (Figure 3). There was no positive result for chromogranin or cytokeratin cocktail, ruling out carcinoma; and no positivity for S100, Melan A, and HMB-45, ruling out melanocytic tumor.

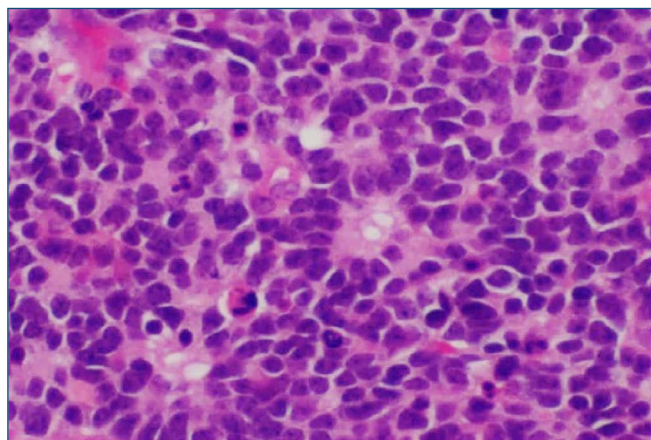


Figure 2. Histological section showing small tumor cells with hyperchromatic nuclei forming Homer Wright rosettes, suggestive of retinoblastoma (hematoxylin and eosin x400).

The diagnosis was unilateral stage E retinoblastoma in adult. After enucleation, a follow-up visit took place every 12 months, for the last 10 years. Right eye fundus test, and brain and orbit MRIs had no findings of OD disease or trilateral retinoblastoma. Genetic testing of the patient or sample was not possible.

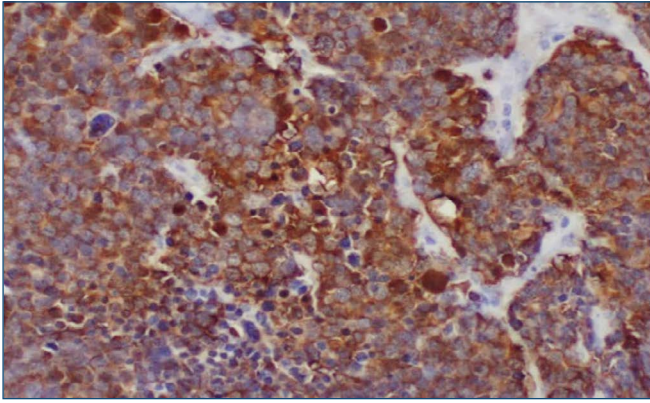


Figure 3. Histological section with immunohistochemical staining and strong and diffuse positivity for neuron-specific enolase (x200).

DISCUSSION

Retinoblastoma is a disease that predominantly affects children, and seldom adults. Clinically it is difficult to differentiate retinoblastomas in adults from amelanotic melanomas, retinal astrocytoma, metastatic tumors, intraocular lymphoma, fungal granuloma or ocular cysticercosis.⁽³⁾

As general rule, adult retinoblastomas are sporadic, unilateral, and progressive, but retinocytomas can be found as residing intraocular masses. Retinoblastomas developing from previous retinocytomas have been described.⁽⁴⁾ It is not clear if adult retinoblastomas are *de novo* retinocytomas or arise from preexisting lesions,⁽⁵⁾ and both cases require a RB1 gene biallelic inactivation, according to the model described by Knudson, in 1971.⁽⁶⁾ There are no previous reports of this patient indicating a history of retinocytoma as malignant tumor predecessor lesion.

The final diagnosis was made using immunohistochemistry tests with highly specific antibodies to identify different retinal cells. In this case, the tumor was highly positive for NSE, confirming the retinoblastoma; however, it is unspecific, since it is also positive for neuroblastomas and Merkel cell tumors, but can be used in combination with H&E stains, which highlighted the Homer-Wright

rosettes found in this case. Negative results for Melan A and HMB-45 ruled out choroidal melanoma.⁽⁷⁾

Delayed diagnosis and invasion of choroidal tissue and the optic nerve are some of the main risk factors for metastasis of retinoblastomas. The case report described did not present any of these and, during the 10-year follow-up, no intraocular lesions in the OD or MRI brain, and orbit lesions have been found. Since no genetic tests were performed, a decision was made to follow-up using long-term imaging, considering 12% to 15% of unilateral cases present the same mutational profile as bilateral cases, predisposing the appearance of second tumors.⁽⁸⁾

Enucleation is the treatment recommended for advanced stages and continues to be the main therapeutic option for most adult retinoblastoma cases reported in the literature⁽⁹⁾. There is little information about long-term follow-up in these patients.

CONCLUSION

Conservative treatment was not possible due to advanced neovascular glaucoma. After enucleation, the patient has remained free of disease, no relapse in the anophthalmic cavity, and no metastasis or suspicious lesions in the healthy eye.

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