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Case study

Medulloepithelioma of the optic disc

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Summary A 6-year-old boy with a history of an amblyopic, occasionally red left eye was found to have a solid white mass overlying the optic disc on dilated ocular fundus examination. Transvitreal endoincisional biopsy of the mass yielded neoplastic tissue consistent with intraocular medulloepithelioma. The eye was removed subsequently because of concern that the tumor may invade the retrobulbar optic nerve. Histopathologic and immunohistochemical analysis of the tumor confirmed nonteratoid medulloepithelioma of the optic disc. The child has been followed up for over 36 months without any signs of orbital tumor recurrence or metastasis.

1. Introduction

Medulloepitheliomas are rare tumors arising from the cells of the primitive neural tube and the medullary plate that are grouped among other primitive neuroectodermal tumors of the brain and spinal cord [1]. In the eye, medulloepitheliomas usually arise from the nonpigmented ciliary epithelium [2] but, on rare occasions, may originate from the iris, retina, or optic nerve [3]. These neoplasms comprise a spectrum from benign to malignant histomorphologically [4]. Invasive intraocular medulloepitheliomas extend out of the eye occasionally via the sclera or optic nerve but rarely metastasize unless neglected or mismanaged. To our knowledge, only 8 other cases of medulloepithelioma of the optic nerve have been reported in the peer-reviewed literature at this time [1,5-11] (Table 1). However, our case is unique in that the tumor was diagnosed by means of cytopathologic study of a tumor specimen obtained by incisional biopsy using a 25-gauge vitrectomy system.

2. Case report

A 6-year-old healthy white boy was evaluated ophthalmologically after falling. He also presented with a complaint of having an amblyopic eye detected during preschool vision screening evaluation. He had a negative family history for eye and systemic disorders. Visual acuity was correctable to 20/20 OD but only to 20/70 OS. Ophthalmic examination of the right eye revealed entirely normal features. Although the

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anterior segment of the left eye was normal, a discrete dull white fundus mass was present overlying the optic disc (Fig. 1A). The tumor measured 6.5 × 5.25 mm in maximal and minimal diameters and 3.5 mm in thickness. It contained several subsurface foci of black pigment and exhibited limited vascularity. B-scan ultrasonography confirmed the solid soft tissue character of the tumor and absence of intralesional calcification (Fig. 1C and D).

A baseline magnetic resonance imaging (MRI) of the orbits and brain was performed to rule out central nervous system (CNS) astrocytic hamartomas, evaluate the orbital optic nerve, and further characterize the epipapillary tumor. The study showed no significant CNS lesions or any retrobulbar expansion of the optic nerve (Fig. 1B). Although these findings helped us to clinically rule out pilocytic astrocytoma of the orbital optic nerve and tuberous sclerosis–related astrocytic hamartoma, the precise nature of the intraocular tumor remained uncertain. Our differential diagnosis included inflammatory granuloma, medulloepithelioma, isolate retinal astrocytoma and adenoma or adenocarcinoma of the retinal pigment epithelium (RPE), and atypical retinoblastoma.

Because a diagnostic consensus based solely on imaging studies was impossible to reach and the possibility that this tumor was a retinoblastoma, tumor sampling for cytologic or histopathologic diagnosis seemed a viable alternative. Choices such as fine-needle aspiration biopsy (FNAB), posterior vitrectomy with endoincisional biopsy, and enucleation were contemplated.

FNAB of the tumor was performed via a transvitreal route but failed to yield a sufficient specimen for cytodiagnosis. An endoincisional biopsy of the tumor was performed immediately after the unsuccessful FNAB using a 25-gauge vitrectomy system and trans pars plana technique. The procedure was entirely uncomplicated. Pathologic evaluation of papanicolaou (Pap)-stained slides of the specimens obtained by endoincisional biopsy disclosed clumps of blue, neuroepithelial cells arranged in cords, and fibrillary material with features suggestive of an embryonal neoplasm consistent with medulloepithelioma (Fig. 2A). The slides were sent to 2 outside consultant ophthalmic pathologists for second opinions, and both concurred with our presumptive pathologic diagnosis.

During the ensuing 2 months, the child experienced slowly worsening vision in the affected eye; however, follow-up ophthalmoscopy showed no significant tumor growth. In view of this progressive visual impairment and because of the concern about possible extraocular extension of the optic disc tumor if left untreated, the child's parents consented to enucleation. This procedure was performed without complications 3 months after the biopsy. Grossly, the tumor appeared as a discreet white epipapillary mass containing scattered clumps of black pigment and small intralesional cavities (Fig. 2B). Histopathologic analysis revealed a neoplastic proliferation of cells resembling the medullary epithelium without heteroplastic elements arranged in cords and tubules of well-polarized epithelium, pseudorosettes, scattered pigment clumps, and cystic spaces.

Table 1 Literature review of cases of medulloepithelioma of the optic nerve

Author	Patient age	Symptoms and signs	Tumor features	Treatment	Adjuvant treatment	Follow-up (survival)
Correa et al (our case)	6 y	Amblyopia OS detected 2 years ago	ON head	Enuc	No	3 y
Takei et al (2007) [6]	~2 y	Proptosis OS for 2 wk	Retro-orbital without cranial extension	Chemo	No	2 y
Chavez et al (2004) [5]	20 mo	Esotropia OD for 4 mo	ON head and sheath to orbital apex	Cran, Enuc + ON resec at optic canal	Chemo	5.5 y
Chidambaram et al (2000) [1]	1.5 y	Proptosis OS 2 mo, ↓ vision and ocular motility, retinal detachment	Retro-orbital with intracranial extension	Resec with complications	None	3 d
Biswas et al (1999) [7]	3 y	Proptosis OS, ↓ eye movements OS 3 mo, NLP	Retro-orbital with intracranial extension	Resec	Rad, 4600 Gy	18 mo
O'Keefe et al (1997) [8]	2 y	Change in iris color + red eye	Intraocular with anterior ON thickening	Enuc + Cran + ON Resec to optic chiasm	None	4 y
Mullaney (1974) [9]	1 y	Inflamed OS, nonreactive pupil, leukocoria	ON tumor (arising from Bergmeister papilla)	Enuc	No	4 mo
Green et al (1974) [10]	6 y	Ocular pain 3-4 d + dilated pupil OD	Not reported	Enuc + Exent + Cran with ON	No	18 mo
Reese (1957) [11]	4.5 y	Drooping left eyelid	Left orbit and optic canal	Enuc + ON res to OC recurrent Exent + res	52 Gy + TEM 1.5 mg	4 mo

Abbreviations: OD indicates right eye; OS, left eye; ON, optic nerve; Chemo, intravenous chemotherapy; Enuc, enucleation; Cran, Craniotomy; Resec, resection; NLP, no light perception; Exent, exenteration, Rad, radiation; TEM, triethylenemelanine.