Melanocytoma of the optic nerve head - a diagnostic dilemma

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Abstract

The clinical features, autofluorescence, B-scan ultrasonography, optical coherence tomography and fluorescein angiography of the lesion were described. Multiple investigation modalities are needed to confirm the benign nature of the lesion. Careful evaluation and follow-up is crucial to avoid misdiagnosis and erroneous management.

Introduction

Melanocytoma of the optic disk is a hamartoma with malignant potential. Sometimes, it may be difficult to distinguish from malignant melanoma. We describe the clinical features, autofluorescence, B-scan ultrasonography, optical coherence tomography and fluorescein angiography of a case of melanocytoma of the optic disk.

Case Report

A 40-year-old Chinese gentleman, presented to our department with right subconjunctival hemorrhage after an episode of vomiting. There was no blurring of vision or any other visual symptoms. At examination, his vision was 1.0 bilaterally. Intraocular pressure was 10 mm Hg (OS) and 12 mm Hg (OD). There was a sectoral subconjunctival hemorrhage at the nasal aspect of his right eye. There were no other remarkable findings on his right eye. Incidentally, there was a light brown lesion at the optic nerve head. The lesion was elevated with feathery margins, involving the inferior and nasal expect of the optic nerve head, extending from the 5:00 to 11:00. Pigmentation was heavier at the inferonasal optic disc margin. The lesion was about 1.5 disc-area in size with the involvement of adjacent choroid. Retinal involvement was less prominent with slight obscuration of the overlying major retinal vessels (Figure 1A). There was neither glial proliferation nor dragging of the retinal vasculature. Lipofuscin deposition, retinal hemorrhages, retinal edema and subretinal fluid were absent. There were no other retinal lesions or ocular melanocytosis. The vitreous was clear. Pupillary light reflexes were brisk and equal bilaterally.

The lesion did not manifest any autofluorescence (Figure 1B). B-scan ultrasonography showed an elevated dome-shaped lesion with an initial tumor thickness of 1.4 mm (Figure 2A). The adjacent choroidal involvement had low internal reflectivity (Figure 2B). Optical coherence tomography (Heidelberg Spectralis® Tracking Laser Tomography; Heidelberg Engineering, Carlsbad, CA, USA) revealed a predominant choroidal involvement with marked displacement of the overlying retina nerve fiber layer (Figure 3). The optic nerve architecture was markedly distorted. Fluorescein angiography revealed fine retinal vasculature on the surface of the tumor; there was no choroidal vasculature supplying the lesion. In the late phase, the hyperfluorescence at the nasal optic disc margin increased size and intensity (Figure 4). Visual field examination (Humphrey, Central 30-2 threshold test, stimulus III) showed a mildly enlarged blind spot. The above features were consistent with melanocytoma of the optic nerve head. The patient is examined periodically. The tumor remains stable; there is no deterioration of visual function or evidence of tumor growth to date.

Discussion

Pigmented tumors of the optic nerve head raised the possibility of malignancy. Primary optic nerve melanoma is extremely rare, whereas secondary invasion of optic nerve by uveal melanoma is uncommon. Lindegaard et al found that the frequency of optic nerve invasion in uveal melanoma varied between 5% and 7%. Increased intraocular pressure and juxtapapillary location of uveal melanoma were risk factors associated with optic nerve invasion.[1] The features of the optic nerve tumor in this patient were consistent with melanocytoma of the optic disk. Nevertheless, the presence of malignancy should be ruled out with a reasonable degree of certainty; due to the aggressive nature of choroidal melanoma and to avoid erroneous enucleation in benign cases.

Melanocytoma of the optic nerve head is a hamartoma consist of large, uniform round or oval heavily pigmented melanocytes that are packed closely. The cells are located among axons in the optic disc, anterior optic nerve and in the peripapillary retina.[2] This accounts for the total absence of autofluorescence. Classically, the appearance of optic disk melanocytoma was described as intensely black thickening of the optic disc with a feathery periphery. Hyperpigmentation may vary from dark brown to black. The degree of pigmentation of the tumor in our patient was lesser than those classically described for a

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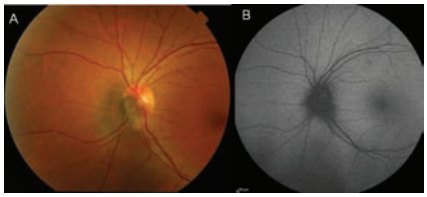

Figure 1. A) Color photo of the left optic disk; B) Absence of autofluorescence.

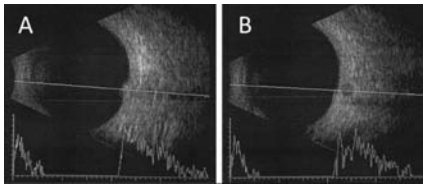

Figure 2. B-scan ultrasonography of the tumor. A) The lesion had a dome-shaped appearance; B) Adjacent choroidal extension of the tumor had low internal reflectivity.