Optic Disc Melanocytoma: A Case Report

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Research Article

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Abstract

Background: Optic disc melanocytoma is a rare tumor of the optic nerve that typically occurs on the surface of the optic disk. Our purpose was to identify a case of optic disc melanocytoma.

Case presentation: A 51-year-old female patient presented with chief complaints of black shadows in the left eye, having normal visual acuity. Fundus examination of the left eye revealed a homogeneous black bulging mass located on the optic disc. The mass demonstrated a hypofluorescence throughout an angiogram at all stages and had differentiated from a malignant melanoma.

Conclusion: The patient presented with black shadows in the left eye. After careful examination, an optic disc melanoma in the left eye was found. Even if it is benign, malignant transformation may occur in the future. Therefore, a follow-up is recommended for a long time.

Introduction

Melanocytoma of the optic disc is a type of melanocytic nevus that occurs primarily in the optic disc, and sometimes, with the involvement of the adjacent choroid and retina^[1]. It is considered to be a congenital and non-genetic lesion, without racial preference, and is usually unilateral to the eye. Its mean age of diagnosis is between 40 and 50 years, with a slightly greater predominance in female patients^[2]. This melanocytoma may cause papilledema, as well as optic nerve damage, and always presents a dark brown or black pattern, being the tumor size of usually around 3 mm^[3]. Melanocytoma of the optic disc is a pigmented benign tumor that is usually considered a quiescent lesion; however, this tumor grows slowly and gradually affects the visual field and vision^[4]. It has been described that in 47-86% of the cases, the adjacent choroid is involved, whereas retinal involvement is described in 30-67% of the cases^[5]. Furthermore, in 25% of the cases, a fundus examination may demonstrate papilledema, and 2% of tumors can progress to a malignant transformation. The tumor may be associated with plasmacytoid retinal detachment, retinal edema, lipid deposition as well as intraretinal hemorrhage. Vitreous pigment dispersion, in some cases, may extend into the anterior segment of the eye. About 75% of patients are asymptomatic, with incidental detection of the tumor. There are several common symptoms associated with this clinical condition including blurred vision, decreased visual acuity, visual perversion, and microvision^[4]. Melanocytoma has been reported in association with ocular melanocytosis, congenital pigment dispersion hypertrophy, choroidal nevus, and retinitis pigmentosa.

Melanocytoma of the optic disc is clinically and histopathologically mistaken with uveal melanoma. The diagnosis of melanocytoma is usually made by examining the optic disc due to characteristic pigmented masses on this disc^[7]. Ancillary tests include ultrasound, fluorescein, and indocyanine green angiography, fundus autofluorescence, and optical coherence tomography (OCT)^[8]. Fundus autofluorescence is a non-invasive imaging method. In rare cases, spontaneous necrosis or progression into a melanoma can occur, resulting in severe vision loss. Although the course of the disease is considered relatively stationary, it is known to increase slightly in 10–15% of the cases. However, increased tumor size and secondary loss of

vision do not correlate with a malignant transformation^[9]. The main biological mechanism, involved in this disease, is the ischemic necrosis of the optic disc and some portions of the retinal nerve fiber layer, caused by the tumor's occupying effect. In early stages, malignant melanoma can mimic melanoma, leading to difficulties in the diagnosis. Considering its rarity and diagnostic difficulties, we report a case of melanoma to describe its clinical features and diagnostic approach.

Case Presentation

A 51-year-old female patient presented with chief complaints of black shadows in the left eye. For further diagnosis and treatment, the patient was referred to the Ophthalmology department, at Qingdao's municipal hospital, on May 31st, 2021.

The ocular examination showed that her best-corrected visual acuity was 0.8 with normal pupillary reactions in both eyes. Slit-lamp biomicroscopy showed normal anterior segments in both eyes, and no abnormalities were found in the systemic examination. Furthermore, a fundus examination of her left eye showed a homogeneous black mass located on the optic disc, occupying the superior one-half, where the vessels in that area were significantly bulged (Figure 1). It was observed that the superotemporal quadrant, adjacent to the retina and the macular fovea, did not reflect light.

The remaining portion of the posterior segment was normal in the left eye, and no abnormality was found in the right eye. Intraocular pressure was measured being 15 mmHg and 17 mmHg for the right and left eye, respectively. Fluorescein angiography of optic disc melanocytoma demonstrated hypofluorescence throughout the angiogram in all stages, and there was no evidence of advanced hyperfluorescence of the mass lesion (Figure 2). A 3D reconstructed image demonstrated that the tumor mass and OCT angiography depicted abundant capillaries at the surface of the tumor (Figures 3A, C, and D). A single hyperreflective band in the local eminence was observed in the OCT, and overlying retinal tissue on the tumor was severely disorganized (Figure 3B). Furthermore, the B-scan ultrasonography of the left eye revealed an elevated small focal lesion with high internal reflection (Figure 4), and a perimetry test of this eye did not reveal any defects in the visual fields (Figure 5).

Altogether, due to its clinical features and the tumor's location, the patient was clinically diagnosed with optic disc melanoma and is currently followed once a month. Since the patient's visual acuity has remained at 0.8 without any changes in lesion size or her visual fields.

Discussion

Optic disc melanocytoma is located on the optic disc and sometimes extends below the sieve plate. Tumor cells are uniform in size, with a pigmented cytoplasm and a low nucleoplasmic ratio. Ruptured specimens show ovoid to round cells with abundant cytoplasm^[1]. The pathogenesis of optic disc melanocytoma remains unknown, but it is thought to be a congenital lesion.

In a retrospective analysis of 115 patients, Shields et al.^[5] found that 99% of patients presented with unilateral lesions and 100% presented with hyperpigmentation. Tumors averaged 2 mm in diameter and 1 mm in thickness. In the present study, the patient showed a similar pattern, with a black mass of approximately 1/2 pupillary distance (PD) in the optic disc of the left eye, protruding from its' surface and obscuring some retinal vessels. The fundus of the right eye showed no significant abnormalities (Figure 1). With a prominent choroidal component, a differential diagnosis of choroidal melanoma, extending into the optic disc, becomes difficult. Salvanos et al.^[6] reported the effectiveness of discriminating optic disc melanoma from choroidal melanoma. Optic disc melanoma shows a completely hypoautofluorescent with clear boundaries, without hyperautofluorescence spots associated with orange pigment.

In most cases, fluorescein angiography of optic disc melanomas shows low fluorescence, however, regarding optic disc edema, dye leakage is common in these lesioned areas. Most melanomas submitted to indocyanine green angiographies also show low fluorescence. In this study, early frames showed a hypofluorescent tumor, and hyperfluorescent optic disc tissue with few vessels, whereas late frames showed dye leakage from the optic disc tissue (Figure 2). Shields et al.^[7] analyzed OCT of 15 optic disc melanocytoma cases, showing hyperreflectivity on the anterior surface of the tumor, and dense posterior shadowing in all cases with an optically cavernous appearance. Okubo et al.^[4] identified hyperreflective spots around central retinal veins and arteries of the optic disc, and perhaps this mechanism may be related to phagocytosis or lipid deposition. In the present case, a single hyperreflective band was detected in the local eminence, and the overlying retinal tissue on the tumor was severely disorganized (Figure 3). Additionally, OCT angiography depicts abundant capillaries at the surface of the tumor.

High-resolution B-scan was used by Gologorsky et al.^[8] to evaluate signs of malignant transformation in benign lesions, including increased size and angiogenesis, and transformation of nodule appearance. In this case, this technic showed a dome-shaped mass lesion over the optic disc with high echogenicity (Figure 4). Melanoma are rarely associated with poor vision, which is related to the compression of the optic nerve, exudative retinal detachment, and malignant transformation of the tumor. Common visual field defects include blind-spot enlargement and nerve fiber bundle defects. It has been defined that the enlargement of the blind-spot is related to the number of tumors extending to the edge of the intervertebral disc. However, in this case, the visual acuity of the patient's left eye was normal and no obvious visual field defects were found (Figure 5).

As referred above, optic disc melanocytoma is a benign tumor that may grow slowly, and rarely transforms into a melanomas, being the rate of malignant transformation of 2%^[3]. There is no established treatment for melanocytoma, and observation is the only clinical tool for its management. However, there are some reports demonstrating that photodynamic therapy and vitreous cavity injections with bevacizumab could be used as a potential treatment of uveal melanoma with choroidal neovascularization^[9]. Tumor necrosis can lead to a misdiagnosis of progressive tumor growth, suggesting its possible transformation into melanoma. In some cases where eyes presented tumor

necrosis, due to temporary ischemic necrosis, vision is restored at the request of the patient according to early reports^[10]. Optic disc melanoma and suspicious conditions are usually treated with enucleation since it has been described that plaque radiation therapy presents lower visual function as well as a higher recurrence rate. Altogether and taking into account the results obtained in this case report, we suggest that patients with optic disc melanocytoma should have a regular ophthalmic examination to monitor the tumor's size, growth rates and prevent its malignant transformation.

Conclusion

The reported case in this study highlights the importance of properly identifying optic disc melanocytoma. Furthermore, whereas these cases do not require treatment a long-term follow-up is needed to exclude and prevent malignant transformations.

Abbreviations

OCT: Optical coherence tomography; PD: pupillary distance

Declarations

Ethics approval and consent to participate

All the subjects have signed written informed consent according to the study protocol approved by the Ethics Committee of Qingdao Municipal Hospital before participation.

Consent for publication

Written informed consent was obtained from the patient according to Ethics Committee Regulations.

Availability of data and materials

The datasets used during the current study are available from the corresponding author on reasonable request.

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Author contributions

Participated in study design: JWY. Contributed to the writing of the manuscript: CXJ. Participated in the topic selection: SDP and ZY.

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Competing interests

The authors declare that they have no competing interests.

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Figures

Figure 1

Fundus photograph showing optic disc melanocytoma of the left eye. A nearly round black mass of around 1/2 PD in size was found on the optic disc of the left eye containing a bulge of approximately 1 mm and a clear edge. This mass protrudes onto the surface of the optic disc and covers part of the retinal vessels. No obvious abnormality in the right fundus was detected.

Figure 2

Fundus fluorescent angiography of the optic disc melanocytoma. (A) The early frames show a hypofluorescent tumor and hyperfluorescent optic disc tissue with few vessels on the tumor. (B) The late frames show dye leakage from optic disc tissue.

Figure 3

Optical Coherence Tomography (OCT) examination of the optic disc melanocytoma. (A) A 3D reconstructed OCT image shows the mass size. (B) A single hyperreflective band was observed in the local eminence, and overlying retinal tissue on the tumor is severely disorganized. (C, D) OCT angiography depicts abundant capillaries at the surface of the tumor.

Figure 4

B-scan ultrasonography axial showing a dome-shaped mass lesion over the optic disc with high echogenicity.

Figure 5

Humphrey visual field analysis. No obvious visual field defects were observed.