

PEER REVIEWED

Optic Nerve Melanocytoma

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Abstract

Optic nerve melanocytoma is a benign variant of melanocytic nevus that is located within or adjacent to the optic nerve. Optic nerve melanocytomas are benign tumors but may cause vision loss and visual field defects. Rarely, these tumors may undergo malignant transformation. We describe a case of optic nerve melanocytoma found on routine examination and describe the demographics, clinical findings, management and prognosis associated with this type of lesion. Thorough evaluation and follow-up are essential to ensure correct diagnosis and appropriate management.

Key Words: *optic nerve melanocytoma, benign ocular tumor, melanocytes, optic nerve neoplasm, optic nerve*

Background

Optic nerve head melanocytoma (ONM) typically appears as black or dark-brown tumors with feathery or “fuzzy” margins located on the optic disc that often extend into the adjacent retina, choroid and vitreous.¹ ONM was first reported in 1907 by Coats, who suspected melanocytomas were benign tumors. In 1962 Zimmerman and Garron described the histopathological characteristics of ONM as round or oval uniform pigmented melanocytes packed closely together. These histological features were consistent with a benign entity, not a malignant neoplasm as was once believed.²⁻⁴ ONM is typically diagnosed clinically based on its characteristic clinical features, although ancillary testing may aid in the diagnosis and prognosis of the lesions. We present a case of optic nerve melanocytoma that was discovered on routine comprehensive eye examination in a 62-year-old patient. We review the clinical presentation, differential diagnosis, ancillary testing, natural history and potential complications of optic nerve melanocytoma. The intended audience is third- and fourth-year optometry students, optometry residents and current practitioners.

Case Description

A 62-year-old African American male presented for a comprehensive eye examination with no visual complaints. His last eye exam, which was three years prior, had been unremarkable. Family ocular history included a parent with glaucoma. Significant medical history included hypertension, post-traumatic stress disorder, osteoarthritis, gout, depression and polysubstance abuse. Medications included acetaminophen PRN for pain, allopurinol for gout, amlodipine for blood pressure, 81 mg aspirin for heart attack/stroke prevention, diphenhydramine for sleep, multivitamin/mineral for nutritional supplementation, pantoprazole for stomach acid, tamsulosin for prostate, topiramate for substance abuse cravings, trazodone for sleep, and ziprasidone for mood. Recent complete blood count, lipid profile, serum glucose, and hemoglobin A1c were within normal ranges. His most recent blood pressure measurement was 134/59 mmHg.

The patient’s best-corrected visual acuities were 20/20 OD and 20/20 OS. Pupils were equally round and reactive to light with no afferent pupillary defect. Extraocular muscle testing was normal. Confrontation

visual fields were full to finger-counting in both eyes, and frequency-doubling screening perimetry with excellent reliability revealed no defects in either eye.

Slit lamp biomicroscopy was unremarkable with trace nuclear sclerotic cataracts noted in both eyes. Intraocular pressure was measured at 18 mmHg in both eyes by Goldmann applanation tonometry. Dilated ophthalmoscopy revealed a black pigmented lesion covering the superior half of the right optic nerve, extending just past the edge of the nerve onto the adjacent retina (**Figure 1**). No significant findings were present in the left eye (**Figure 2**). Color fundus photography was performed for baseline documentation.



Figure 1. Fundus photograph of the right optic nerve.
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Figure 2. Fundus photograph of the left optic nerve.
[Click to enlarge](#)



Figure 3. Heidelberg Spectralis retinal nerve fiber layer scans and analysis OU. [Click to enlarge](#)



Figure 4. Heidelberg Spectralis near-infrared photograph and vertical spectral domain OCT section through the right optic nerve. [Click to enlarge](#)



Figure 5. Heidelberg Spectralis near-infrared photograph and horizontal spectral domain OCT section through the right optic nerve. [Click to enlarge](#)



Figure 6. Humphrey visual field test (central 30-2 threshold) in the left eye.
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Figure 7. Humphrey visual field test (central 30-2 threshold) in the right eye. [Click to enlarge](#)

The patient returned a few weeks later for baseline optical coherence tomography (OCT) imaging (**Figures 3-5**) and automated Humphrey central 30-2 threshold visual field testing. The retinal nerve fiber layer (RNFL) was robust in both eyes, with global values of 119 μm in the right eye and 114 μm in the left eye. The superior quadrants (location of the ONM OD) did not differ significantly between eyes with values of 143 μm OD and 138 μm OS.

The Humphrey visual field test was moderately reliable in the right eye and reliable in the left eye. Visual field in both eyes was essentially clear (**Figures 6-7**). The patient was advised of the findings and instructed to return for follow-up in six months to ensure stability and annually thereafter unless he experienced changes in vision or new ocular symptoms.

Education Guidelines

Learning objectives

1. Understand the typical presentation of optic nerve melanocytoma
2. Understand the clinical findings associated with optic nerve melanocytomas and differential diagnoses
3. How to use ancillary testing to aid in diagnosis and prognosis of optic nerve melanocytoma and potential complications

Key concepts

1. Visual complications may arise from optic nerve melanocytomas even though the tumors are benign
2. Critical thinking in differentiating optic nerve melanocytomas from other ocular tumors
3. Managing patients with optic nerve melanocytomas

Discussion points

1. Do optic nerve melanocytomas affect vision?
2. Do optic nerve melanocytomas affect pupil response?
3. Do optic nerve melanocytomas cause visual field defects and if so what kind?
4. What differential diagnoses should be considered in cases of suspected optic nerve melanocytoma?
5. What ancillary testing can aid in the diagnosis of optic nerve melanocytoma?
6. What is the appropriate management for optic nerve melanocytoma?
7. What is the natural history and prognosis for optic nerve melanocytoma?

Literature review

Optic nerve melanocytomas are benign tumors, with equal incidence among all races.^{1,2} It is a longstanding misconception that ONM is more common in darker pigmented individuals.^{1,5} The average age at diagnosis is 50 years, and there appears to be a slight female predilection, with one study reporting women having a 2:1 likelihood of having ONM.¹ Optic nerve melanocytomas are unilateral, and in rare instances have been reported to occur bilaterally in children, which suggests a congenital etiology.⁵⁻⁶ ONM has no systemic associations but may be associated with intracranial meningiomas.¹

Discussion

Teaching instructions: Participants should read each question and consider how they would respond. Next, they should read the information provided in the text. Participants may work together in small groups or individually, either in real time or as part of a homework assignment. Learning objectives are to be assessed by comparing participants' responses with the information provided. This case may also be presented as a PowerPoint presentation detailing the case description, learning objectives, key concepts, literature review and discussion points.

Do optic nerve melanocytomas affect vision?

In most cases, ONM does not cause significant vision loss.^{1,4} Lee et al. reported that 93% of patients with ONM had vision of 20/40 or better.⁷ Up to 26% of optic nerve melanocytomas may cause mild visual impairment, usually as a result of disc edema, retinal edema or subretinal fluid involving the fovea.^{1,4,8-13} Other potential causes of vision loss include compression of the optic nerve, tumor necrosis, juxtapapillary choroidal neovascularization, central retinal vein occlusion or malignant transformation.^{1,4,8-13} Vision loss may be reversible in some cases.⁶ Severe vision loss is rare and should be looked upon with close scrutiny as it may be evidence of malignant change.³

Do optic nerve melanocytomas affect pupil response?

An afferent pupillary defect (APD) is observed in 10-30% of all ONM.^{1,6} Shields et al. reported that an APD may occur despite normal visual acuity.¹ Conversely, Lee et al. reported that an APD was present only in patients with vision equal to or worse than 20/50.^{4,7} Afferent pupillary defects are likely the result of compression of the optic nerve fibers by the melanocytic cells.¹

Do optic nerve melanocytomas cause visual field defects, and if so what kind?

Visual field defects are commonly reported with ONM and include enlarged blind spot and nerve fiber bundle defects.^{1,4} The most commonly reported defect is enlargement of the blind spot, which is believed to be directly related to the amount of tumor extension and compression of the optic nerve axons.^{1,4} Nerve fiber bundle defects include nasal steps and arcuate defects.¹⁴ Ninety percent of all optic nerve melanocytomas are associated with abnormal visual field findings; thus, it is important to establish baseline visual fields at time of diagnosis.¹ Visual field testing may help detect ONM enlargement or malignant transformation.⁵

What differential diagnoses should be considered in cases of suspected optic nerve melanocytoma?

Differential diagnoses for ONM include choroidal nevus, juxtapapillary choroidal melanoma, metastatic melanoma to the optic nerve, and retinal pigment epithelium (RPE) hyperplasia of the disc. Optic nerve melanocytomas are believed to be variants of choroidal nevi that are located on the optic nerve.^{1,19} Choroidal nevi are typically flat or minimally elevated and tend to be located juxtapapillary instead of overlying the disc as optic nerve melanocytomas do.¹ Choroidal melanomas may occur on the optic nerve and can be extremely difficult to distinguish from ONM. Choroidal melanomas tend to be lighter in color compared to optic nerve melanocytomas and may have associated subretinal fluid or overlying lipofuscin, which are not seen in ONM. Metastatic melanomas often present clinically as unilateral or bilateral optic nerve edema due to diffuse infiltration of the optic nerve, and a distinct darkly pigmented lesion is not observed.^{1,19} Unlike patients with optic nerve melanocytomas that remain asymptomatic, patients with metastatic optic nerve melanomas typically complain of acute pain, reduced vision and diplopia.²⁰ Metastatic choroidal lesions, which appear as unilateral or bilateral creamy white or pale yellow elevated lesions, often accompany optic nerve metastasis.²⁰ RPE hyperplasia at the optic nerve margin may present similarly to ONM; however, the margins are typically more irregular and not as feathery or fuzzy as in ONM. A history of trauma or inflammation may be elicited from the patient, and there be accompanying chorioretinal scarring.

What ancillary testing can aid in the diagnosis of optic nerve melanocytoma?

Ancillary testing is not necessary for diagnosing ONM as the diagnosis is made by assessing the clinical features. However, ancillary tests can aid in diagnosis and be useful in monitoring the lesions and determining prognosis.

Spectral domain OCT through the ONM typically shows an elevated, dome-shaped lesion on the anterior surface of the optic nerve and characteristic posterior shadowing due to loss of light transmission through the pigmented tumor.¹⁵⁻¹⁷ Like ONM, choroidal nevi also appear as dome-shaped lesions with deep shadowing on OCT imaging. However, unlike ONM, choriocapillaris compression overlying the nevus, photoreceptor loss and RPE atrophy are also noted on OCT imaging of choroidal nevi.²¹ Choroidal melanomas display similar OCT features to choroidal nevi, including deep optical shadowing and choriocapillaris compression. A distinguishable difference between the two is the presence of “shaggy” photoreceptors of choroidal melanomas due to the subretinal fluid that typically accompanies choroidal melanomas but not ONM.²¹ Choroidal metastases display irregular “lumpy bumpy” lesions unlike the smooth dome-shaped lesions of ONM, choroidal nevi and melanomas.²¹ Choroidal metastases also display RPE abnormalities and compression of the choriocapillaris.²¹

OCT angiography may be useful in identifying abnormal retinal vasculature on the tumor surface and in the peripapillary region, which has been described as a risk for tumor growth.^{7,16} OCT is valuable in measuring tumor thickness for monitoring, as well as in detecting possible complications of ONM such as subretinal fluid.⁵

Fundus autofluorescence (FAF) of ONM reveals a characteristic hypoautofluorescence, while the remaining retina demonstrates isoautofluorescence.¹⁵ Similarly, fluorescein angiography (FA) reveals hypofluorescence throughout.^{1,18} Neither FA nor FAF are much help with ONM diagnosis.

B-scan ultrasonography is an important diagnostic and prognostic tool. Melanocytomas greater than 0.5 mm in elevation may be visualized with B-scan. ONM may be monitored over time with B-scan for change and malignant transformation. Gologorsky et al. observed 90% of optic nerve melanocytomas to have medium to high reflectivity on B-scan, 62% to be dome-shaped, and 28% to be mildly elevated.¹⁹ Choroidal melanomas are also typically dome shaped on B-scan ultrasonography but exhibit low to medium reflectivity, whereas choroidal metastases are more irregular in shape and exhibit high reflectivity.²² Choroidal nevi appear as thin choroidal masses with tapering margins that blend into the normal choroid with moderate reflectivity on B-scan ultrasonography.²³

What is the appropriate management for optic nerve melanocytoma?

No treatment is indicated for optic nerve melanocytoma. Patients should be monitored annually with dilated fundus examination and fundus photography.

What is the natural history and prognosis for optic nerve melanocytoma?

Although ONM tumors are benign, they have a 1-2% risk of conversion to malignancy.^{1,6} ONM may exhibit slow growth over time, but this does not always indicate malignant change.¹ Conversely, rapid tumor growth or necrosis with severe vision loss is highly suspicious for malignant transformation, and the patient should be promptly referred to an ocular oncologist.⁴ The main predictive factor for growth is tumor thickness greater than 1.5 mm at initial diagnosis.¹

Conclusion

Optic nerve melanocytomas are darkly pigmented benign tumors located partially or completely within the optic nerve head. Despite being considered benign tumors, they may cause vision loss and/or visual field defects due to various complications. Rarely, these lesions may convert to malignancy; thus, annual monitoring for change is recommended.

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