Optic Disc Melanocytoma: A Rare Case of Hamartoma

Mariam Shamim[1]*, M. Asghar Rajani[2], Mudassir Hilal[3]

[1]Department of Ophthalmology, Prevention of Blindness (POB), Eye Hospital Karachi, Pakistan
[2]Prevention of Blindness (POB), Eye Hospital Karachi, Pakistan
[3]Prevention of Blindness (POB), Eye Hospital Karachi, Pakistan

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***Correspondence to**
Mariam Shamim
Prevention of Blindness (POB), Eye Hospital Karachi, Pakistan
Email mariamshamim@hotmail.com

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ABSTRACT

Optic disc melanocytoma is a rare benign tumor arising from melanocytes within the optic disc. We present a case of 42-year-old female with an incidental finding on left eye optic disc. Fundoscopic examination revealed a darkly pigmented mass on optic disc with feathery margins. OCT demonstrated dome-shaped mass with a brightly reflective anterior surface and posterior optical shadow. Optical coherence tomography angiography (OCT-A) revealed intact retinal vasculature with dilated major retinal vein. Optic disc melanocytoma is typically managed conservatively due to its benign nature, however long term follow-up is essential to monitor for potential complication or growth. This case highlights the importance of recognizing optic disc melanocytoma as a differential diagnosis in patient presenting with optic disc lesion, emphasizing the role of imaging modalities in diagnosis and management decision.

Keywords: Benign Tumor, Optic disc tumor, BSCAN ultrasound, Optical Coherence Tomography, Autoflourescence.

INTRODUCTION

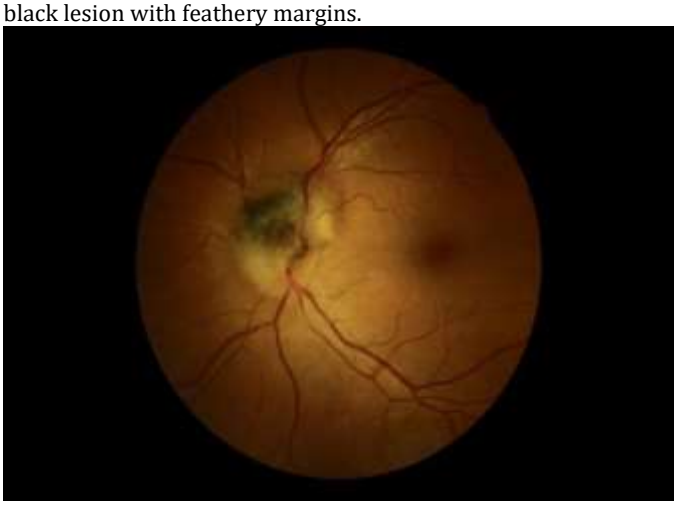
Optic disc melanocytoma is a hamartoma.[1] It is a benign, stationary, densely pigmented tumor that develops wherever uveal melanocytes are present, and most commonly occurs on or near the optic disc. They are usually unilateral, and there is a 1-2% chance of malignant transformation[12]. Although rare, melanocytoma can cause local complications such as disc edema, sub retinal fluid, focal hemorrhage, and retinal vein obstruction. Occasionally it is difficult to differentiate melanocytoma from malignant melanoma. Considering its rarity and diagnostic dilemma, we report a case of optic disc melanocytoma defining its clinical features, B-scan ultrasonography, fundus autofluorescence, visual field, optical coherence tomography, and optical coherence tomography angiography of melanocytoma of the optic disc.

CASE REPORT

A 42-year-old woman visited our hospital with a complaint of decreased near vision. Her vision was 6/6 bilaterally and N6 with +1.00. Her intraocular pressures were 12 (OD) and 14 mmHg (OS). Anterior segments of both eyes were unremarkable on Slit Lamp Bio-microscopic examination. The pupillary reactions were normal. Left eye fundus exam showed a dark brown to black elevated lesion with feathery margins at the superior border of the optic disc, it was approximately one-disc diameter in size and was obscuring the optic disc as shown in Figure1.

There was no evidence of any associated sub-retinal fluid, orange pigment, or disc edema. Fundus examination of the right eye was normal.

Figure-1: Fundus photograph of left eye showing a dark brown to black lesion with feathery margins.



On further investigations, B-scan ultrasonography of the left eye revealed a hyperechoic small dome-shaped lesion. (Figure-2) Fundus autofluorescence showed the lesion to be hypoautofluorescent. Spectral-domain optical coherence tomography (SD-OCT) (Heidelberg Spectralis®; Heidelberg Engineering) imaging showed a dome-shaped mass with a brightly reflective anterior surface and posterior optical shadow. (Figure-3) Optical coherence tomography angiography (OCT- A) revealed intact retinal vasculature with dilated major retinal veins (Figure-4).

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Figure-2: B-scan ultrasonography showing dome shape lesion.



Figure-3: Spectral-domain optical coherence tomography (SD-OCT) of the lesion showing increased reflectivity at its anterior margins and a dark, optically empty space at its posterior margin due to tumor shadowing.

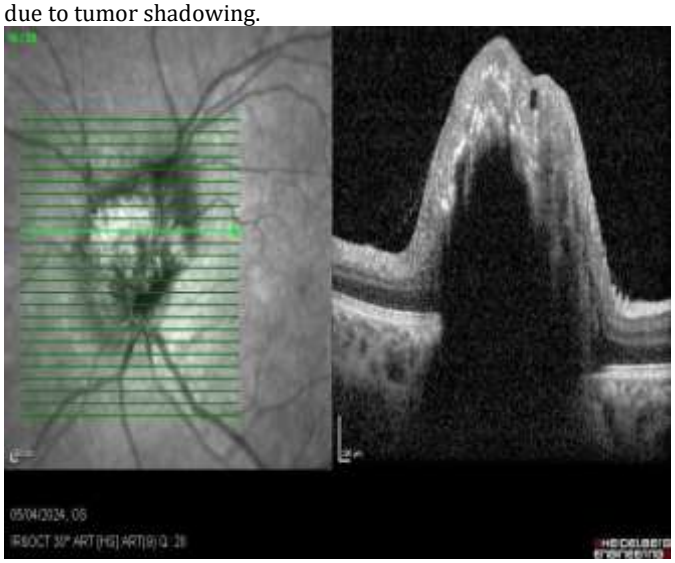
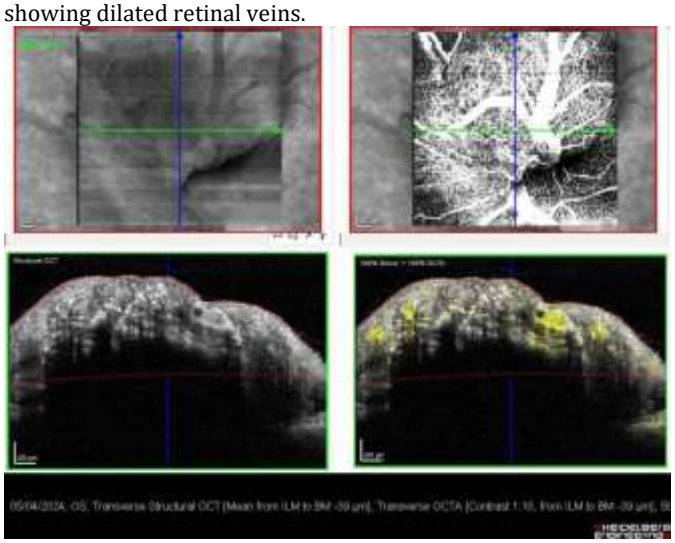


Figure-4: Optical coherence tomography angiography (OCT- A) showing dilated retinal veins.



DISCUSSION

Although, optic nerve melanoma is exceedingly rare [1,2,] pigmented tumors of the optic nerve head significantly raise the likelihood of malignancy. It is therefore essential to rule out malignancy with reasonable certainty due to the aggressive nature of the melanoma and to prevent unnecessary enucleation in benign cases.

Melanocytoma of the optic nerve head is a hamartoma that consists of uniform, large, round, or oval densely packed and heavily pigmented melanocytes. These cells are situated in the optic disc, peripapillary retina and anterior optic nerve among the axons.[2] This explains the total absence of autofluorescence. Frequently, the appearance of optic disc melanocytoma is characterized by a deeply black lesion on the optic disc with feathery margins.[14] Hyperpigmentation may vary from dark brown to black. Rarely, melanocytoma can present as an amelanotic lesion, which later develops pigmentation.[3] The presence of melanocytoma usually does not result in significant visual impairment.[13] It is often diagnosed incidentally. Visual loss in optic disc melanocytoma could be caused by compression on the optic nerve, juxtapapillary choroidal neovascularization, retinal exudation, tumor necrosis, central or branch retinal vein occlusion, and malignant transformation.[4,18]

Imaging modalities like B-scan ultrasonography, optical coherence tomography, fundus autofluorescence, and optical coherence tomography angiography can help in understanding the tumor morphology and confirming the diagnosis.[5,15]

On SD-OCT, a melanocytoma appears as an elevated dome-shaped mass that originates from the optic disc. It has increased reflectivity at its anterior margins and a dark, optically empty space at its posterior margin due to tumor shadowing.[16] The internal limiting membrane is disrupted with disorganization of overlying and adjacent retinal layers.[20] In cases of melanoma, SD-OCT shows a highly reflective choroidal layer with thickening of the RPE- Bruch membrane and the presence of sub-retinal and intraretinal fluid.[5]

B-scan ultrasonography reveals an elevated dome shape lesion with medium to high internal reflectivity. Gologorsky, Daniel, et al in their study found out 89%of melanocytoma had medium to high internal reflectivity. [11] Features suggestive of a high risk of malignant transformation are increased tumor thickness, low internal reflectivity, nodular appearance, and rapid increase in size.[6]

SW-AF is a non-invasive, dye-free test., while the remaining retina shows iso- autofluorescence.[19] SW-autofluorescence is useful, non-invasive and dye free test that is useful in differentiating between melanocytoma and choroidal melanoma. Optic nerve melanocytoma appears as a hypo-autofluorescent lesion, while choroidal melanoma shows discrete and bright hyper-autofluorescence due to the presence of overlying intracellular lipofuscin.[7]

OCTA is useful for noninvasively documenting choroidal neovascularization (CNV) associated with melanocytoma. It can also aid in detecting secondary issues such as retinal vein or artery occlusions. In our case, the patient's retinal veins were more dilated in the affected eye, likely due to limited outflow obstruction.[8,17] The optic nerve tumor in our patient exhibited features consistent with melanocytoma of the optic disc. The absence of lipofuscin deposition, retinal hemorrhages, and subretinal fluid argued against the diagnosis of malignant melanoma in our patient. Additionally, the results of the ancillary testing further supported our diagnosis.

CONCLUSION

Optic nerve melanocytoma is a benign tumor with potential for

malignant transformation, local complications, and visual loss. Careful evaluation and testing are crucial for a correct diagnosis, and patients need lifelong observation.

Ethical Approval
Ethical approval for this case report was obtained from the relevant institutional ethics review committee, in accordance with institutional guidelines.

Data Availability
All relevant clinical information supporting this case report is included in the manuscript.

Author Contributions
Conceptualization: Mariam Shamim, M. Asghar Rajani
Clinical Evaluation and Data Collection: Mariam Shamim, Mudassir Hilal
Investigation: Mariam Shamim, M. Asghar Rajani, Mudassir Hilal
Writing – Original Draft: Mariam Shamim
Writing – Review and Editing: M. Asghar Rajani, Mudassir Hilal
Supervision: M. Asghar Rajani

Informed Consent
Written informed consent was obtained from the patient for publication of this case report and the accompanying clinical details and ophthalmic images. The patient's confidentiality and anonymity have been maintained.

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Conflict of Interest
No.

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Declaration of AI-Assisted Language Editing
Grammarly AI was used for language and grammar correction, while Gemini AI was used for figure generation.

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