

## CASE REPORT

## OPTIC DISC MELANOCYTOMA; A RARE ENTITY

**Jamaluddin Mughal, Anum Javed, Owais Arshad, Javeria Nasir, Mohammad Hanif Chatni, Mohammad Usman**

Ophthalmology Department, Patel Hospital, Karachi-Pakistan

A young male presented with complaints of disturbance of vision and floaters for the past five years with best corrected visual acuity of 6/6 in both eyes. Ophthalmic examination of the anterior segment was normal in both eyes. There was a relative afferent papillary defect in the right eye. Fundoscopic examination of the left eye showed a raised, 3-disc diameter (DD) dark brown lesion arising from the optic disc. Ophthalmic investigations were performed including fundus photos, A-scan, B-scan, Auto-fluorescence, OCT, and FFA. Findings were consistent with those of optic disc melanocytoma. The patient is now kept on close three-monthly follow-up. To the best of our knowledge, there are no published reports from Pakistan so far.

**Keywords:** Optic Disc Melanocytoma; Melanocytic Lesion; Choroidal Melanoma

**Citation:** Mughal J, Javed A, Arshad O, Nasir J, Chatni MH, Usman M. Optic Disc Melanocytoma; A rare entity. J Ayub Med Coll Abbottabad 2020;32(4):580–2.

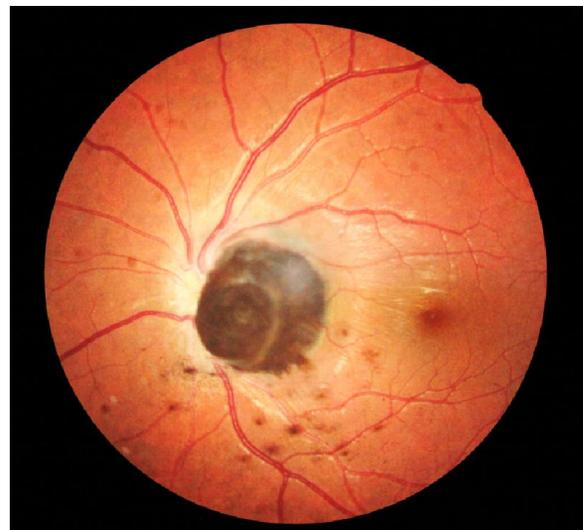
## INTRODUCTION

Intraocular melanocytic lesions can range from small, flat nevi to enlarged and elevated lesions with malignant potential. Distinguishing and diagnosing these can be a challenging task owing to the similar presentation of benign and malignant lesions.<sup>1</sup> Optic Disc Melanocytoma is a primary, rare, benign, unilateral, highly pigmented tumor of the optic disc with leafy margins. In the past, patients with intraocular melanocytic lesions have been enucleated based on suspicion of being melanomas. Although ODM has a very low potential for malignant transformation, it has been previously reported.<sup>2</sup> We report a case diagnosed as optic disc melanocytoma based on history and relevant ophthalmic investigations which included fundus photos, A/B scan ultrasonography, and OCT, and systemic radiological examination.

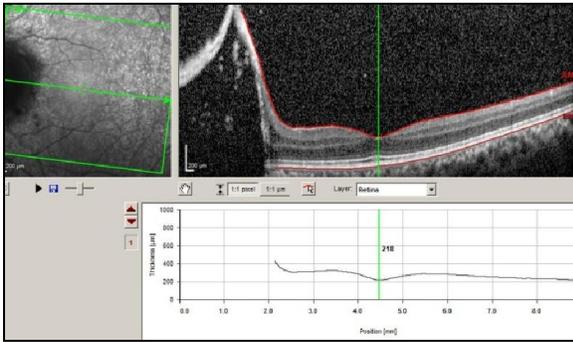
## CASE REPORT

A 24-year-old male came with complaints of floaters and blurring of vision in the left eye for the past five years. It was gradual in onset and not associated with pain or flashes of light. His best corrected visual acuity (BCVA) was 6/6 in both eyes. Slit lamp examination showed a relative afferent papillary defect in the right eye, a normal anterior segment in both the eyes. The intra-ocular pressure was within normal limits. Dilated fundus examination was normal in the right eye. In the left eye, the fundus showed a 3DD lobulated, elevated deeply pigmented lesion, measuring 3×3 mm in diameter, having feathery margins, and protruding from the disc. It was likely emerging from the lamina cribrosa. There were very fine, small, multiple, scattered pigments around the disc, inferio-nasally (Figure-1). He had no other co-morbid. Systemic examination was unremarkable. We kept primary choroidal melanoma in our differential diagnosis and investigated accordingly.

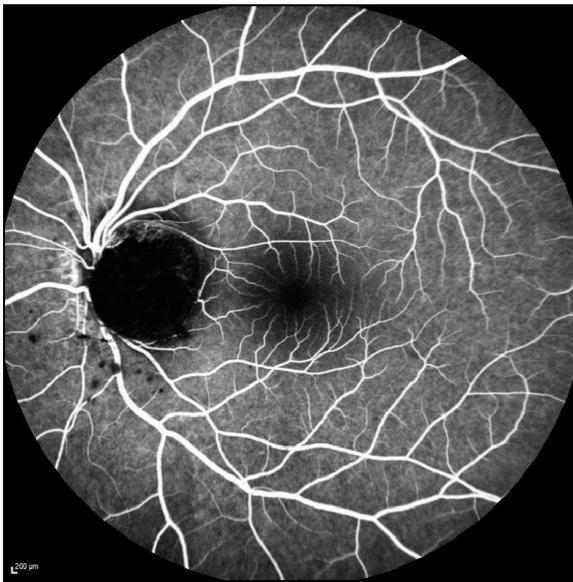
Fundus photography of the right eye was normal. A-scan showed medium to high reflectivity in the left eye. B-scan showed the lesion to be of 3×3 mm with no acoustic hollowness. The OCT showed a sloped, bright reflective surface of the tumour along with retinal distortion and posterior optical shadowing with no sub-retinal fluid (Figure-2). The visual field was measured showing normal field in the right eye and only central island of vision twenty to thirty degrees, in the affected left eye. Fundus fluorescein angiography illustrated diffuse blocked hypo-fluorescence in all phases in the area covered by the pigmented lesion, with diffuse hyper-fluorescence at the temporal rim of the lesion. (Figure-3). Ultrasound abdomen and chest x-rays were normal. A bone scan revealed no signs of metastasis.



**Figure-1: Left eye fundus showing a globular, elevated deeply pigmented lesion with very fine, small, multiple, scattered pigments around the disc, inferio-nasally**



**Figure-2: SD-OCT findings of the left eye showing hyper-reflective elevated area**



**Figure-3: Left eye FFA showing diffuse blocked hypo-fluorescence in late phase**

## DISCUSSION

Optic disc melanocytoma (ODM) is a rare, benign, unilateral, dark brown to the black lesion that occurs either on or beside the optic nerve head arising from the melanocytes. Before Zimmerman labelled these lesions as benign, they were considered to be malignant, and affected eyes were mostly enucleated as melanomas.<sup>3</sup>

Optic nerve head pigmented lesions have a broad spectrum including choroidal melanomas, nevi, RPE hyperplasia along with primary metastatic melanoma of the optic nerve and hamartomas. It is of utmost importance to differentiate between these lesions, based on the clinician's skills and the relevant investigations. Multiple studies have concluded a 2% chance of malignant transformation of optic disk melanocytoma.<sup>4</sup>

The level of visual impairment associated with ODMs is variable as studied by Attiku and other

colleagues.<sup>5</sup> Usually, the vision in affected patients is stable but in rare cases, multiple causes of declining vision have been identified including optic neuropathy, tumor necrosis, choroidal neovascularization, retinal traction, epi-retinal membrane, central retinal vein occlusion, or malignant transformation. If progressive visual deterioration is found, malignant transformation should be suspected because of the optic nerve ischemia occurring secondary to spontaneous tumor necrosis.<sup>6</sup> In our patient, a visual field defect was noted in the left eye with the only central island of vision present. These findings were consistent with Al-Rashaed *et al.*<sup>7</sup> The left eye showed medium to high internal reflectivity. No acoustic hollowness was observed. A recent study by Andres showed similar findings, in which the A/B scans of ODM showed moderate to increased reflectivity.<sup>8</sup>

Spectral-Domain Optical Coherence Tomography visualizes the retinal and choroidal infrastructure. OCT findings of retinal and choroidal lesions have been described earlier.<sup>9</sup> Our findings of OCT included a sloped, bright, hyper-reflective elevation along the tumour margin with peripheral retinal disturbances which are consistent with the work of Guerra *et al.*<sup>10</sup>

The fundus auto-fluorescence of our patient showed a hyper-fluorescent, sharp, bright image with well-defined borders, due to the presence of densely packed melanocytes as observed by Zhang *et al.*<sup>11</sup>

Fundus Fluorescein Angiography illustrated diffuse blocked hypo-fluorescence in all phases in the area covered by the pigmented lesion, with diffuse hyper fluorescence at the temporal rim of the lesion as seen by Shields and fellows.<sup>12</sup> He is now on the regular 3-monthly follow-up to keep a track of the lesion.

## CONCLUSION

Although ODMs are rare benign entities with seldom visual impairment these tumours should be kept on sequential follow-up by ophthalmologists with serial imaging for any disturbances, due to mass-effect or malignant transformation.

**Patient's Consent:** Informed consent was taken from the patient about this case report.

**Conflict of Interest:** All authors declare no conflict of interest.

## REFERENCES

1. Harbour JW, Paez-Escamilla M, Cai L, Walter SD, Augsburger JJ, Correa ZM. Are risk factors for growth of choroidal nevi associated with malignant transformation? Assessment with a validated genomic biomarker. *Am J Ophthalmol* 2019;197:168–79.

2. Pereira A, Thomas S, Yadav NK, Venkatesh R. Multicolor imaging of optic disc melanocytoma. *Indian J Ophthalmol* 2019;67(12):2056–7.
3. Mansour AM, Zimmerman LO, La Piana FG, Beauchamp GR. Clinicopathological findings in a growing optic nerve melanocytoma. *Br J Ophthalmol* 1989;73(6):410–5.
4. Baartman BJ, Ahmad B, Srivastava S, Jones S, Singh AD. Melanocytoma or juxtapapillary melanoma?. *Retinal Cases and Brief Reports*. 2019 Jan 1;13(1):15-7.
5. Attiku Y, Rishi P, Bassi S. Coexisting Optic Disc Melanocytoma and Pituitary Adenoma. *Ocul Oncol Pathol* 2019;5(5):319–22.
6. Herwig-Carl MC, Loeffler KU, Grossniklaus HE. Melanocytoma of the conjunctiva: clinicopathologic features of three cases. *Ocul Oncol Pathol* 2019;5(4):290–7.
7. Al-Rashaed S, Abboud EB, Nowilaty SR. Characteristics of optic disc melanocytomas presenting with visual dysfunction. *Middle East Afr J Ophthalmol* 2010;17(3):242–5.
8. Kita Y, Holló G, Murai A, Kita R, Hirakata A. Optical coherence tomography angiography findings of an optic disc melanocytoma in a glaucoma eye. *Int Ophthalmol* 2019;39(3):677–82.
9. Carnevali A, Querques L, Zucchiatti I, Scorgia V, Bandello F, Querques G. Optical coherence tomography angiography features in melanocytoma of the optic nerve. *Ophthalmic Surg Lasers Imaging Retina* 2017;48(4):364–6.
10. Guerra RLL, Marback EF, Silva IS, Maia Jr OD, Marback RL. Autofluorescência e tomografia de coerência óptica de domínio espectral do melanocitoma do disco óptico. *Arq Bras Oftalmol* 2014;77(6):400–2.
11. Zhang P, Hui YN, Xu WQ, Zhang ZF, Wang HY, Sun DJ, *et al*. Infrared autofluorescence, short-wave autofluorescence and spectral-domain optical coherence tomography of optic disk melanocytomas. *Int J Ophthalmol* 2016;9(5):713–6.
12. Shields JA, Demirci H, Mashayekhi A, Eagle Jr RC, Shields CL. Melanocytoma of the optic disk: A review. *Indian J Ophthalmol* 2019;67(12):1949–58.

Submitted: February 17, 2020

Revised: --

Accepted: July 31, 2020

**Address for Correspondence:**

**Anum Javed**, Ophthalmology Department, Patel Hospital, Karachi-Pakistan

**Email:** anum.javed1989@gmail.com