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A Case Series of Optic Nerve Head Melanocytoma from a Tertiary Care Hospital in Rural Tamil Nadu

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Abstract

Optic Nerve Head Melanocytoma (ONHM) is a rare benign pigmented condition manifested due to the presence of Uveal melanocytes more predominantly on or just adjacent to the optic nerve head. In spite of having a rare possibility for being malignant, it has previously been misdiagnosed to be a malignant melanoma that resulted in the execution of various enucleations. We report a case series of three patient's with Optic Nerve Head Melanocytoma, all noted as incidental findings on routine examination. Though benign, we would like to stress on advising a lifelong follow-up in these patients.

Keywords: Malignant Melanoma, Melanocytoma, Optic Disc Melanocytoma, Uveal Tract Tumours.

Introduction

Optic Nerve Head Melanocytoma is a rare benign pigmented condition involving the optic nerve head. Zimmerman in 1962 described this as a benign hamartomatous tumor of the melanocyte (1). Malignant transformation is noted in 2 out of 115 patients in a study by Shields *et al.* (2). Enlargement in the size of the lesion is noted in 10 to 15 percent of the cases. Differentiating it from Malignant Melanoma is very important to prevent unnecessary enucleation of the eye and to prevent psychological stress to the patient. Malignant transformation, although rare, has been reported in approximately 2% of cases. Given the potential for growth and rare malignant transformation, lifelong follow-up is essential for patients with Optic Nerve Head Melanocytoma (ONHM). Our case series highlights the importance of identifying this rare condition, emphasizing the need for thorough diagnostic evaluation and long-term monitoring to ensure optimal patient outcomes.

Case Series

CASE-1: A 30-year-old male presented to the ophthalmology department for routine examination. On ocular examination, his visual acuity was noted to be 6/18 and 6/12 respectively. Best corrected visual acuity in both the eyes was 6/6 with correction of -2.00 Dioptre sphere in the Right eye and - 1.5 Dioptre sphere in the Left eye with normal pupillary reactions. The intraocular pressure noted in the right and left eye were 16- and 12-mm hg respectively. On slit lamp examination the anterior segments were normal. Dilated fundus examination of the Right eye showed the media was clear (Figure 1). A dark brown to black elevated mass lesion of about ¾th disc diameter size with feathery margins, obscuring the disc margin was noted. Diagnosis of optic nerve head melanocytoma was made. No significant evidence of sub retinal fluid, retinal edema, disc edema or serous retinal detachment could be traced. The left eye appeared to be normal (Figure 2). The visual field examination was normal. No evidence of sub retinal fluid noted. On B scan examination of the right eye showed a dot like elevation with medium internal reflectivity (Figure 3). Optical coherence tomography was normal (Figure 4). The Color fundus photo was taken for documentation. Patient examination on 6 months follow up showed a similar finding (Figure 5). The patient is now on regular follow up.

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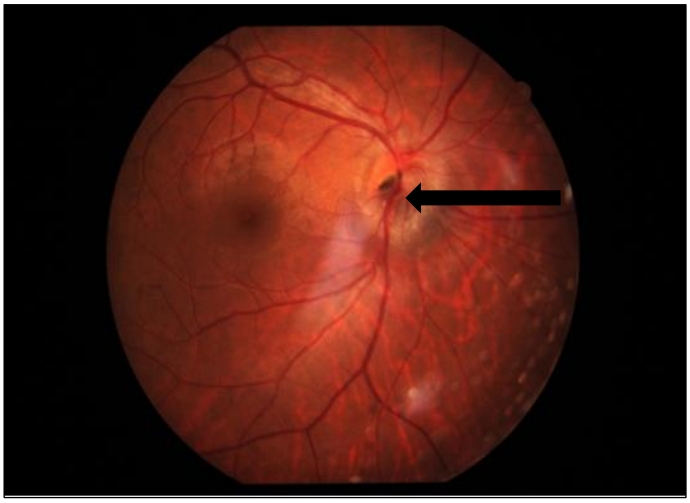

Figure 1: Colour Fundus Photograph of Right Eye Showed Clear Media with a Dark Brown to Black Elevated Mass Lesion of About ¾th Disc Diameter Size with Feathery Margin (Indicated by Arrow)

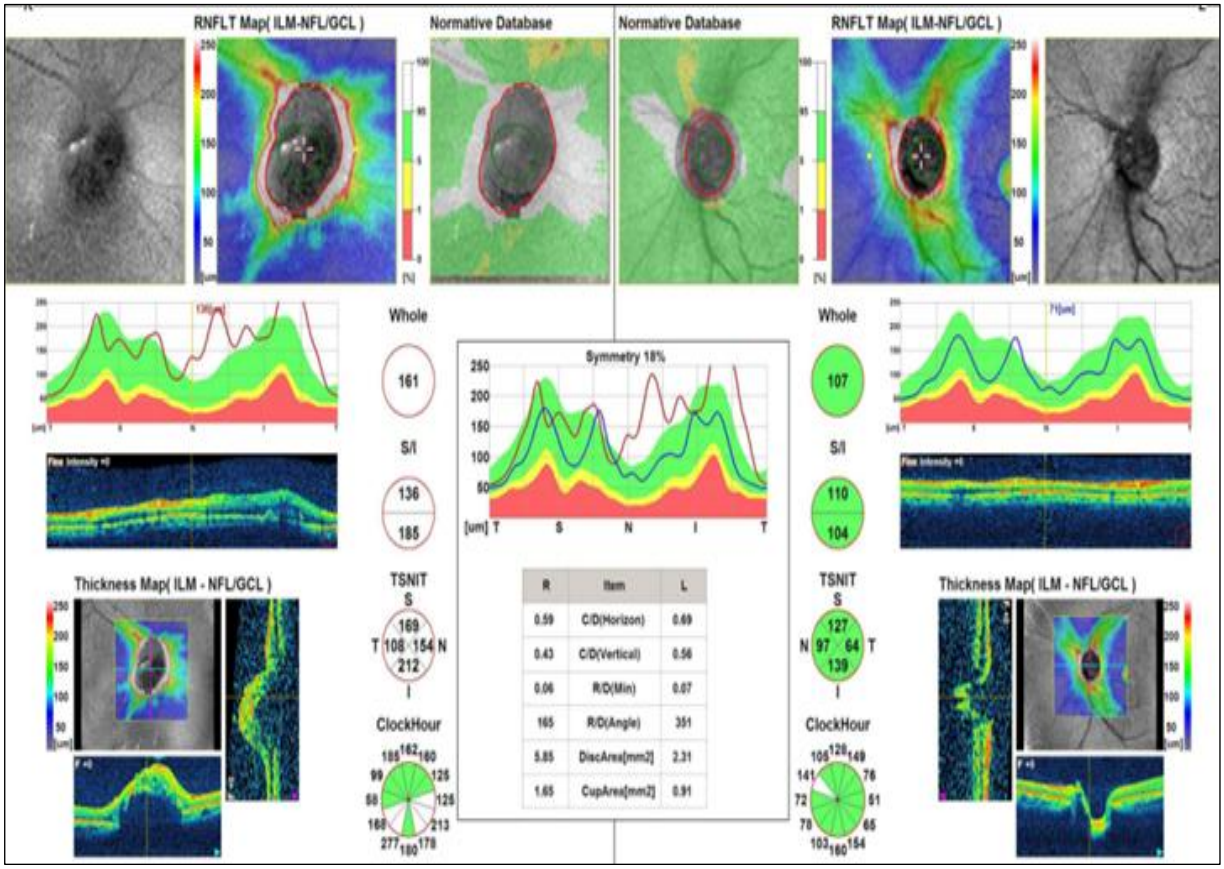

Figure 2: Normal Visual Field of Case 1

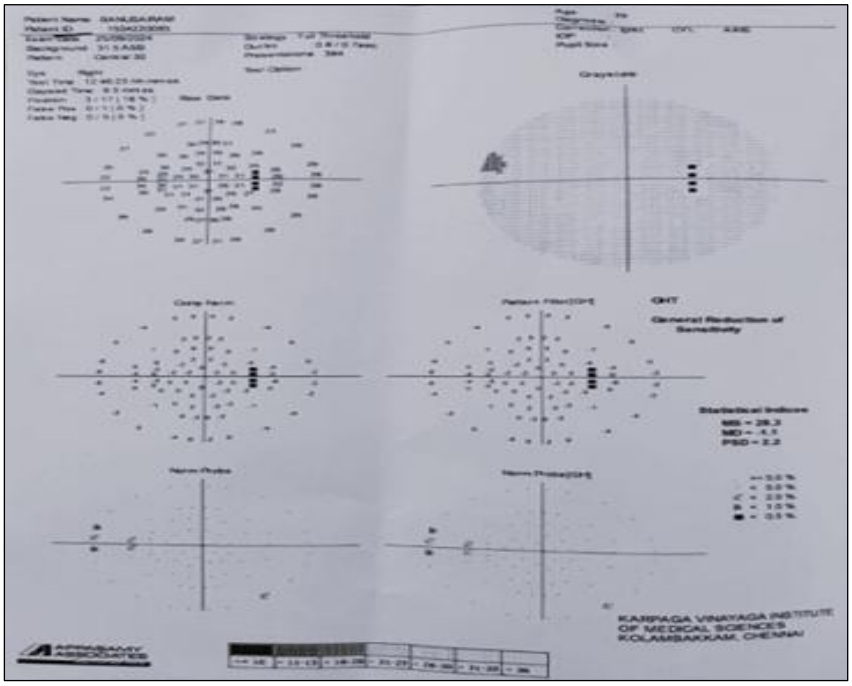

Figure 3: B Scan of Case 1 Shows a Dot like Elevation with Medium Internal Reflectivity (Indicated by Arrow)

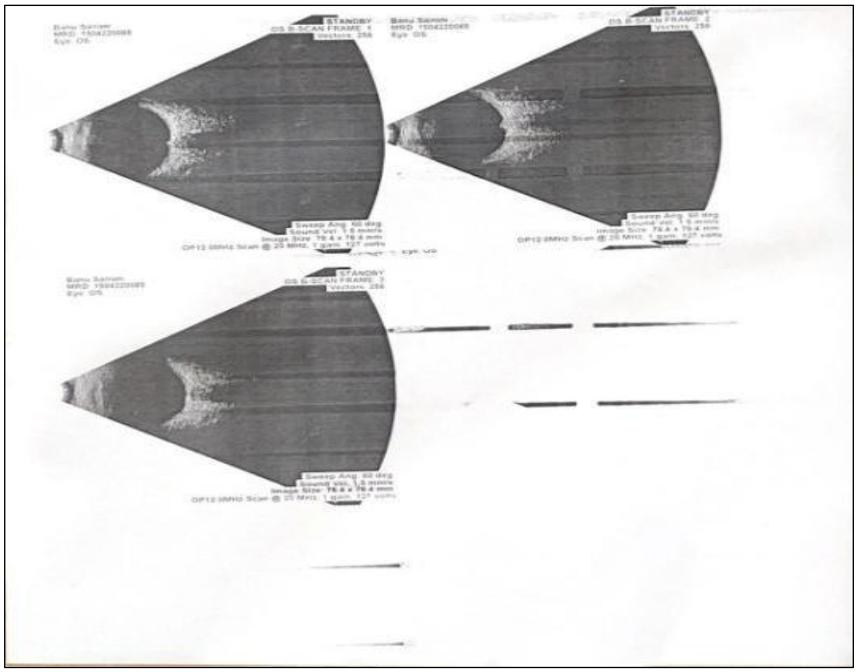

Figure 4: Normal OCT of Case 1

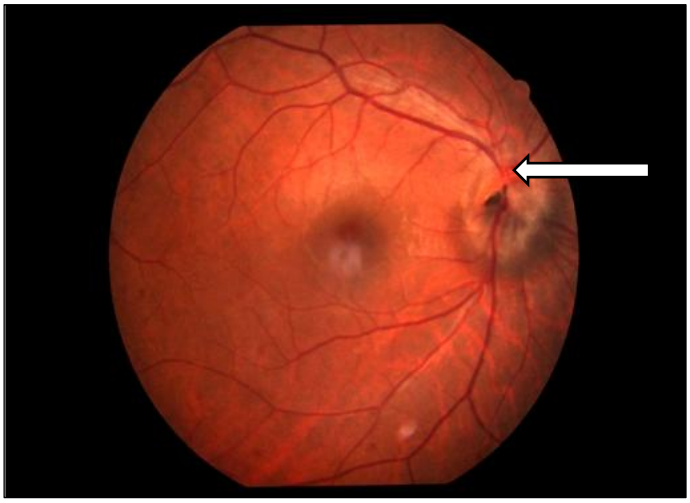

Figure 5: Follow-Up Image of Case 1 after 6 Months

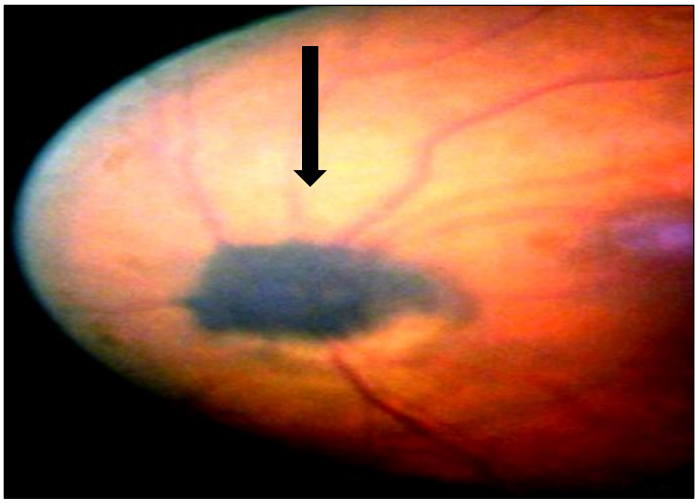

Figure 6: Fundus Picture of Case 2- Right Eye; Clear Media with Dark Brown to Black Elevated Lesion of Half Disc Area Size with Feathery Margins (Indicated by Arrow)

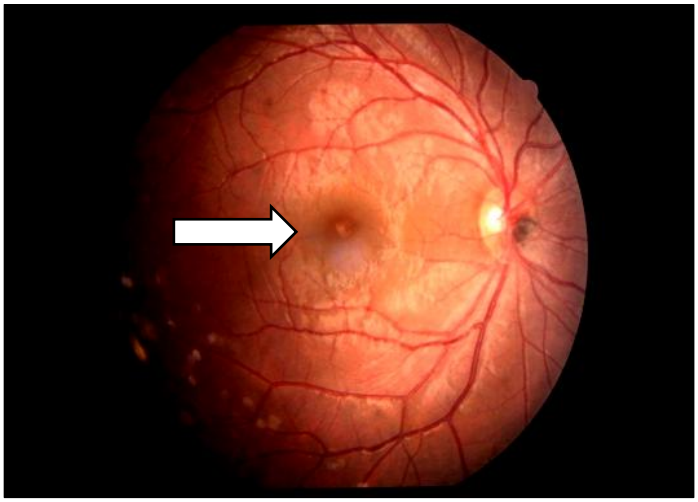

Figure 7: Colour Fundus Picture of Case 3- Left Eye; Hazy Media due to Cataract with a Black Slightly Elevated Lesion with Feathery Margin at the Optic Nerve Head (Indicated by Arrow)

CASE-2: A 52-year-old male presented for routine examination. He did not have any systemic comorbidity. His best corrected visual acuity was 6/6 in both the eyes with the normal pupillary reactions. The intraocular pressures noted were within normal limits. On slit lamp examination the

anterior segment was normal. On dilated fundus examination, the Right eye showed clear media with dark brown to black elevated lesion of half disc area size with feathery margins suggesting the optic disc melanocytoma (Figure 6). Left eye appeared to be normal. Macula was normal. Foveal reflex was noted. Visual field examination and B Scan was not done. Patient was advised to have a regular examination but was lost to follow up.

CASE-3: A 55-year-old female presented to the ophthalmology department with the complaints of diminished vision in both the eyes. On examination, the best corrected visual acuity in both eyes was 6/12. The intraocular pressures were within normal limits. On slit lamp examination both eyes had immature cataract. Dilated fundus examination of both the eyes showed hazy media due to cataract. There was a black slightly elevated lesion with feathery margin at the optic nerve head of the left eye which is suggestive of Optic Nerve Head Melanocytoma (Figure 7). There was no evidence of peri papillary edema or vascularity at the lesion. The right eye appeared to be normal. Visual field examination showed enlargement of blind spots.

Discussion

Optic nerve head melanocytoma also known as magno cellular nevus of optic nerve (3). Though it is a benign condition it has to be differentiated from uveal malignant melanoma. The mean age at diagnosis of this condition is about 50 years. One of our patients is 30 years old but there is a case report of melanocytoma in patients from 1- 90 years. In our case series, in all the three cases there was no reduction in visual acuity due to the lesion. Poor visual acuity may happen owing to the optic nerve compression, exudative retinal detachment, central retinal vein occlusion, tumor necrosis as well as the malignant transformation of the tumour (4). Involvement of the adjacent retina was noted in about 30% of cases according to the study by De potter *et al.*(5). Regarding the tumour malignancy, vascularity is spotted out to be a cardinal marker which is often present in intraocular malignancy. However, Optic nerve head melanocytoma has always been contemplated as an avascular tumour on the basis of Fundus Fluorescein Angiography and Indocyanine Green Angiography that do not always exhibit surface or the intra-lesion vascularity largely because of the occurrence of dense tumour pigmentation. A study by Vishal Raval *et al.,* showed that Optical Coherence Tomography-Angiography may disclose the presence of surface tumour vascularisation in Radial Peripapillary Capillary slab and also the intrinsic vascularity in the choroidal slab as well (6). A rare Ocular association of ONHM with peri papillary hyperreflective ovoid mass-like structures reported by Fubin Wang in 2023. Spectral domain optical coherence tomography showed an elevated tumour mass arising from the optic disc with increased reflectivity. Peri papillary ovoid mass-like structures appeared ovoid in shape in optical coherence tomography (7). The prime feature of malignant melanoma is the presence of a vascular mass lesion with an orange pigmentation, suggesting the lipofuscin which is hyper auto fluorescent (1). The defects with the visual field are typically characterized by the enlarged blind spot depending on the extent to which the tumor is elongated further on the optic disc margin and there may also be the nerve bundle defect owing to the optic nerve compression. One of our patients showed a normal field and one patient visual field examination was not done. Another patient had an enlarged blind spot. A study by Usui *et al.,* in their case series, had enlargement of blind spots in 70% cases (8).

Table 1: Distinguishing Features of Optic Nerve Sheath Meningioma, Melanocytoma, and Optic Nerve Head Drusen (2, 9, 10)

Feature	**Optic Nerve Sheath Meningioma (ONSM)**	**Optic Nerve Head Melanocytoma (ONHM)**	**Optic Nerve Head Drusen (ONHD)**
Etiology	Benign tumor arising from arachnoid meningothelial cells.	Benign pigmented tumor of melanocytic origin	Hyaline bodies within the optic nerve head
Onset and Progression	Gradual, insidious visual loss over months to years.	Slow-growing, stable, or minimal progression	May cause gradual visual loss; typically, asymptomatic

Visual Acuity	Progressive decline in vision, often unilateral.	Mild or no visual decline, can be incidental	Typically, normal unless compressing adjacent nerve fibers
Visual Field Defects	Altitudinal defects, generalized constriction, or enlarged blind spots.	Mild field defects; usually arcuate or paracentral	Inferior or nasal arcuate scotoma (if any)
Optic Disc Appearance	Optic atrophy, Opto ciliary shunt vessels.	Pigmented mass on or near the optic disc	Buried or visible calcified drusen on the optic disc
Pain	Typically, painless.	Painless	Painless
Imaging: MRI	Tubular enlargement of the optic nerve sheath, "tram-track" sign on axial views Coronal view:"doughnut" sign.	Hyperintense on T1-weighted imaging, often iso- or hypointense on T2	Small hypointense spots at the optic nerve head
Imaging: Ultrasound (B-scan)	Optic nerve sheath thickening, sometimes with calcifications.	Often not detected unless causing mass effect	High reflectivity corresponding to calcified drusen
CT scan	Calcifications may be present, optic nerve thickening.	May show a pigmented mass; no calcifications	Calcified drusen visible as hyperdense spots
Fluorescein Angiography	Normal or mild hyper fluorescence.	Minimal or no leakage	Hyper fluroscence with smooth margin FAF - Hyper autofluorescence of drusen
Diagnosis Confirmation	Biopsy rarely needed due to typical imaging features.	Clinical observation, biopsy rarely needed	B-scan ultrasound and autofluorescence are confirmatory
Treatment	Observation, radiotherapy, or surgical intervention in progressive cases.	Observation, no treatment unless causing visual loss	No treatment necessary unless causing visual field defects

Some distinguishing features of ONSM, ONHM, and ONHD are listed in Table 1. Histopathological examination plays a crucial role in distinguishing Optic Nerve Head Melanocytoma (ONHM) from other pigmentary lesions in the optic nerve head. ONHM is characterized by a benign proliferation of melanocytes, typically exhibiting dense melanin pigmentation and a lack of cellular atypia or mitotic activity (1). In contrast, malignant melanoma of the optic nerve head shows cellular pleomorphism, nuclear atypia, and increased mitotic activity (11). Optic nerve sheath meningioma, another differential diagnosis, is marked by meningeal proliferation and the presence of psammoma bodies (12). Conversely, optic nerve head drusen exhibit calcified deposits and hyaline degeneration, without melanocytic proliferation (13). The absence of necrosis, vascular invasion, and inflammatory response further supports the benign nature of ONHM (14). Accurate histopathological diagnosis is essential for guiding appropriate management and predicting patient outcomes. Ultrasound B scans may be employed as a diagnostic tool for its documentation purposes. Previous research in the field suggests that 20 MHz high-resolution Ultrasound B scan be best utilized for the identification as well as the routine follow up of the

patients who present with ONHM with high-risk manifestation for growth and it can further hamper the possibility of a fallacious prognosis. Melanocytoma in B-scan Ultrasonography may appear in a dome-shaped configuration with medium to high internal reflectivity (15). Two important features that are to be carefully observed are features which may suggest malignant melanoma and also features of local ocular complications. Juxta papillary involvement in malignant melanoma of the choroid can mimic ONHM but it may be difficult to differentiate two conditions. A study by Phillpotts *et al.* showed that ONHM is noted more in African- Asian population but uveal melanoma is uncommon in these populations (16). In case of malignant melanoma of choroid visual loss progressive increase in size of the lesion are more commonly seen. Sudden visual loss in ONHM can occur due to ischemic necrosis of the tumour. ONHM usually noted over the optic nerve head without juxtapapillary choroidal involvement. Where as in malignant melanoma, juxtapapillary papillary choroidal involvement is noted. Study by Shields JA *et al.,* showed that about 8 % patients with ONHM had rare ocular associations like optic disc hypoplasia, retinitis pigmentosa and congenital hypertrophy of the retinal pigment epithelium and rare systemic association like neurofibromatosis and basal cell carcinoma (2). Fundus Fluorescein Angiography are not of much use because of the pigmentation of the lesion which can cause blocked fluorescence. Increased vascularity of the lesion is suggestive of malignant melanoma. CT and MRI may not be helpful in differentiating but documentation and measurement of the lesion and extension of the tumor can be identified with these modalities of imaging. The second important feature to be noted while examination of patient with ONHM are the presence of local complications like serous retinal detachment/sub retinal fluid, haemorrhage, vitreous seeds, and central retinal vein occlusion. All these can cause decrease in vision in these patients. The long-term management of ONHM emphasizes regular monitoring for tumour growth, preservation of visual function, and timely intervention for potential complications, typically involving biannual follow-ups (every 6-12 months), comprehensive visual assessments (visual acuity and field testing), imaging documentation (fundus photography, OCT imaging), selective use of ultrasound and MRI as needed (11, 17, 18).

Conclusion

ONHM is a rare and benign condition which has malignant transformation potential. During the course of the disease process it may also precipitate visual impairment. This affects the quality of life index. Also due to the risk of malignancy in the long term it is crucial to have an early detection and regular follow up. We report this case series to suggest the importance of lifelong follow up and close monitoring for malignant transformation and local complications.

Abbreviations

Nil.

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Author Contributions

Each author contributed equally.

Conflict of Interest

The authors declare no conflict of interest.

Ethics Approval

The patient has given consent for the publication and uses the images.

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