# Optic disc melanocytoma: A case report.

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**Abstract:**

Melanocytoma or magnocellular nevus is a distinctive type of nevus that may originate on optic nerve head, choroid, ciliary body, iris. Melanocytoma of the optic disc is a pigmented lesion, occurring on the optic nerve head and often extends into the peripapillary retina and choroid. It may be sometimes difficult to distinguish between a melanocytoma and a malignant melanoma in the early stage. Although these tumors may grow slowly and invade surrounding ocular tissues, behaviourally they are benign. Ultrasonography is usually not helpful and fluorescein angiography can also often be misleading. Some unfortunate patients get enucleated due to lack of investigational support. We hereby present a case where diagnosis could be confirmed only on histopathological report of the optic nerve section following enucleation.

**Key words**: Intraocular tumor, Magnocellular nevus, Melanoma.

## Introduction:

Melanocytoma of the optic disc is a tumor that arises from melanocytes and is a variant of the melanocytic nevus. Melanocytes are found in uvea and occasionally lamina cribrosa. It is typically a unilateral, stationary and classically asymptomatic lesion which can turn malignant with sudden fall in visual acuity associated with vascular changes, necrosis, hematoma which fortunately are rare. Juxtapapillary malignant melanoma of the choroid may involve the optic disc by direct continuity, or by metastasis, if distant. Primary optic disc malignant melanoma is rare. Malignant melanoma at times may mimic a melanocytoma thereby causing diagnostic difficulty in the early stage [1] .

Ophthalmoscopic features include dark brown or black elevated lesion over optic disc generally located in inferotemporal quadrant with extension over retina and choroid. The choroidal component looks like a juxtapapillary choroidal nevus, retinal component is usually black with feathery margin. Size is typically small with average 2mm in diameter and 1mm thickness. Associated findings may be optic nerve edema, retinal edema, retinal exudates, subretinal fluid and vitreous seeding [2].

Besides ophthalmoscopic examination, some ancillary tests are needed to arrive at a diagnosis. Ultrasonography shows elevated margin of optic disc with mild to moderate

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internal reflectivity showing avascularity. Increased tumor thickness, presence of intrinsic vascularization, and nodular (elevated) configuration are the three parameters used to stratify lesions into low-risk or high-risk for malignant transformation[3].

Visual fields show enlargement of blind spot due to extension past the disc margin and retinal nerve fiber bundle defects from compression at optic disc [4]. Fundus fluorescein angiography shows hypo fluorescent lesion in all phases because the cells are deeply pigmented and closely compact with relatively avascularity. It can be associated with optic disc edema or subretinal fluid. Leakage may signify active lesion with chances of malignancy [5]. Optical Coherence Tomography (OCT) features of melanocytoma of the optic disc are appearance of a thin, delicate, echogenic line delineating the anterior aspect of the melanocytoma associated with an abrupt and complete shadowing behind, obscuring all details of the optic nerve and adjacent retina [6]. Magnetic resonance imaging (MRI) helps to determine periorbital and intracranial extension. A sudden increase in size may alert for malignant potential [7]. The final diagnosis is obtained after histopathological analysis. We hereby present a case where diagnosis was confirmed on histopathological report of optic nerve section following enucleation.

**Case report:**

A female patient aged 49 years came to our tertiary care center with chief complain of left eye sudden, painless, diminution of vision for 3 days, not associated with any other ocular or systemic complaints. She had consulted some practitioner where she was diagnosed optic disc neuritis and referred to a higher center for management. She had a history of using spectacles for five years for distance and near vision. According to her reports, her corrected visual acuity in left eye was 6/60 with +2.00D spherical which had dropped down to no perception of light and right eye was 6/6 without correction. Intraocular pressures (measured by Perkin's Tonometer) were 14mm of Hg in right eye and 12 mm of Hg in left eye. Slit lamp examination revealed normal anterior segment in both eyes except for a relative afferent pupillary defect in left eye. In left eye, color vision was defective but in right eye it was normal. Gonioscopy (done by Goldman 4 mirror) revealed Shaffer grade 3 in both eyes but multiple goniosynechia in left eye. Fundus examination by indirect ophthalmoscope revealed blurring of disc margin with circumferential disc edema and black pigment over inferonasal quadrant extending to peripapillary region, macular edema with macular folds in left eye. (Image 1) The fundus of right eye was within normal limits.

Left eye ultrasonography revealed a 2.5 mm elevation at the optic disc with no evidence of other abnormality (Image 2). MRI of brain and left eye showed 2.7 x 2 mm sized button like abnormal signal intensity lesion at optic disc extending 1.9 mm into optic nerve which appeared hyperintense on T1WI and hypointense on T2/STIR images. The lesion showed enhancement on post contrast studies (Image 3). OCT of left eye revealed a dome shaped elevation at the optic nerve head with shadowing behind obscuring optic disc and retinal layer details (Image 4).

**Image 1: Fundus of left eye showing optic disc melanocytoma**

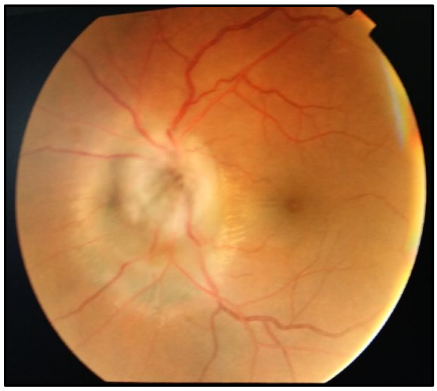


**Image 2: Ultrasonography scan showing elevated optic disc margin with anechoic vitreous cavity in left eye.**

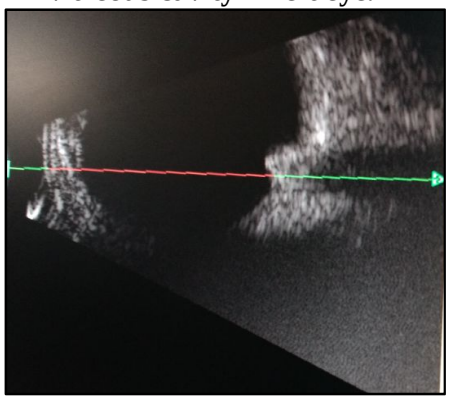
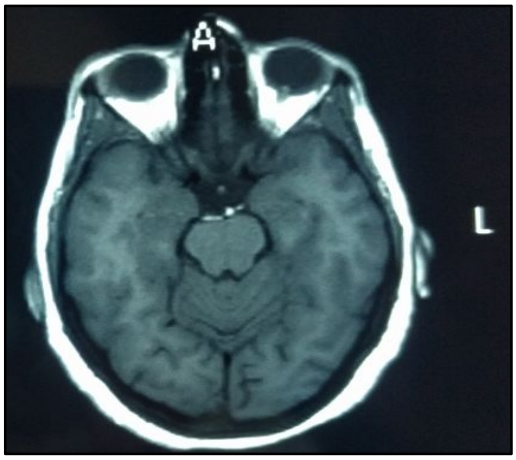


**Image 3: MRI of brain with left orbit Hyperintense lesion on T1, Hypointense lesion on flair T2**



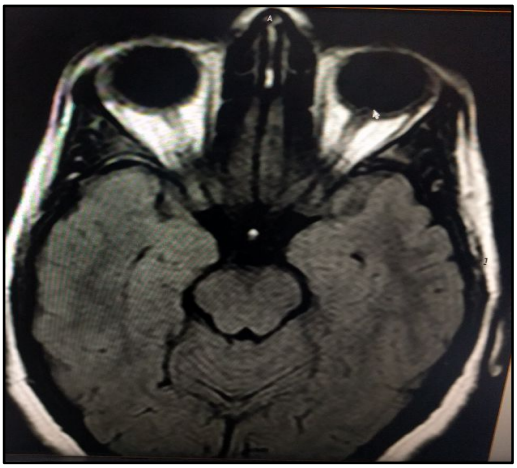



**Image 4: Frequency domain Optical coherence tomography of left eye optic nerve head**

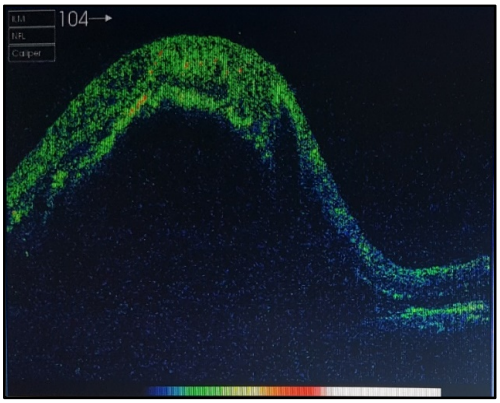
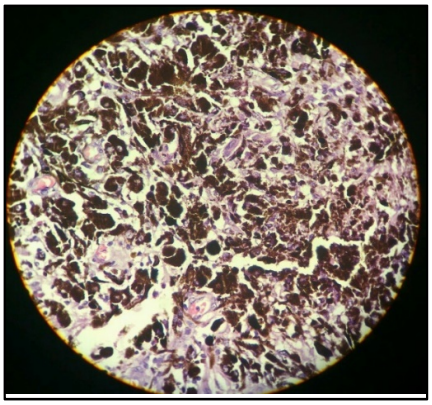



**Image 5: Microscopic view of melanocytes**



Other systemic investigations were within normal limits. The patient was sent to our affiliated cancer hospital where details were reviewed and patient reassessed and the optic disc lesion was suspected to be of malignant potential due to sudden loss of vision. Left eye enucleation was advised. The surgery was performed by the onco-ophthalmologist under

local anesthesia. The left eyeball with optic nerve section of 1.8 cm length was removed. The eyeball was sent for histopathological evaluation which showed a black colored thickened area at optic disc measuring 0.2x0.2x0.2cm$^3$ on gross examination. Microscopic examination of the optic nerve cut end revealed melanocytoma of optic disc showing proliferation of heavily pigmented melanocytes. On melanin bleach, the pigmentation disappeared. The melanocytes were round to polygonal in size with eccentrically situated small vesicular nucleus with inconspicuous nucleoli. Pleomorphism and mitosis was absent. Rest of the optic nerve was free of melanocytic cells which indicate localized non-infiltrating lesion. (Image 5, 6a, 6b, 7a, 7b).

**Image 6: Melanocytes proliferating near normal nerve fiber layer of retina.**

**(A) Before melanin bleach.** **(B) After melanin bleach.**

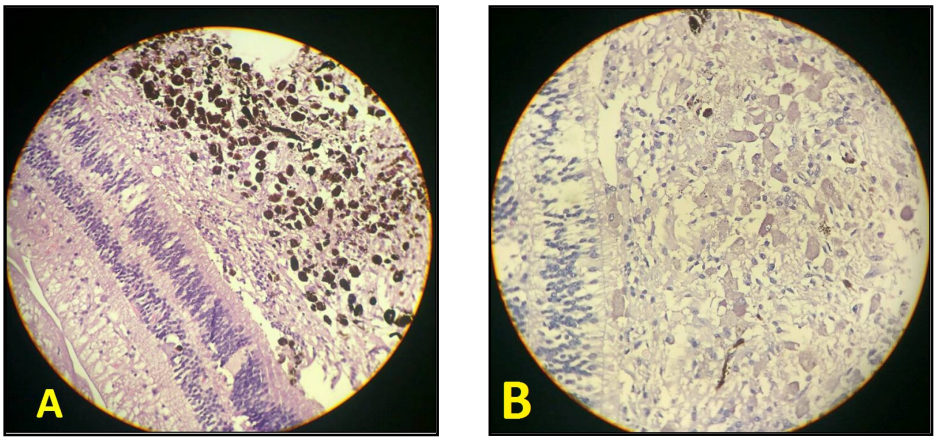
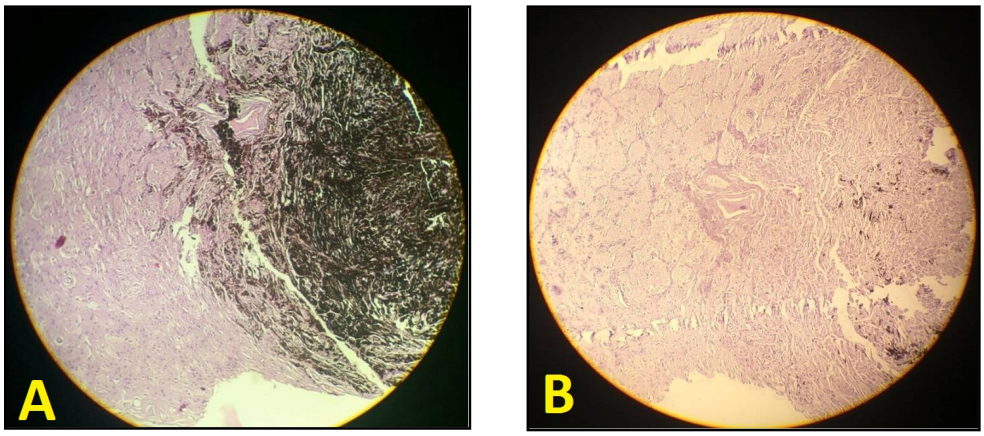



**Image 7: Melanocytes at optic disc with black pigmentation**

**(A) Before melanin bleach.** **(B) After melanin bleach.**




**Discussion:**

It was believed that such pigmented lesions on optic disc were malignant neoplasms until Zimmerman in 1962, declared it to be a benign tumor and coined the term optic disc melanocytoma [1]. He described it as a benign intraocular lesion composed of a uniform array of deeply pigmented, plump, polyhedral nevus cells. With a mean age of presentation of 50 years, females, dark skinned races and Mediterranean races are affected more commonly. The incidence of malignant transformation is 1-2% [1].When a pigmented fundus lesion is

associated with severe visual loss, it is tempting to attribute the visual impairment to malignant transformation, as happened in our case. However, well-documented cases of malignant transformation of optic disc melanocytoma are rare. It is advisable to wait and observe for growth and identify various changes pointing towards a malignant nature. A study by Shields et al of 115 cases of optic nerve head melanocytomas indicates that a significant percentage of the melanocytomas grow if followed long-term (Kaplan-Meier estimate of 32% growth at 10 years) [8]. Two lesions in their study underwent malignant transformation during the follow-up period.

The highest elevation on ultrasonography of any lesion in a study by Gologorsky et al. was 2.6 mm and the largest basal diameter was 4.5 mm, whereas melanomas tend to be larger and more elevated [3]. In our patient, the lesion was 2.5 mm elevated as seen in ultrasonography.

MRI appearance of melanocytoma and uveal melanoma is similar which hence does not aid in diagnosis. Fundus fluorescein angiography is helpful in delineating the vascular nature of the lesion which can show preponderance towards malignancy and can also assess if any vascular obstruction could be the possible cause of vision loss. This unfortunately could not be done in our patient due to her management plan by the oncologists.

Although the visual loss is usually permanent, it has been rarely known to improve after a necrotic episode. [9] Nonetheless, there have been several reported diagnostic dilemmas, including presumed melanocytomas later found to be melanomas. [10] In our case we suspected melanoma which after enucleation and histopathological evaluation was diagnosed as melanocytoma. Histopathological assessment is necessary for definitive confirmation of the nature of the tumor [11].

**Conclusion:**

An unknown and previously undocumented pigmented lesion of the optic disc and peripapillary area is a cause of serious concern. However, knowledge of the specific ophthalmoscopic features of an optic disc melanocytoma can help not only to diagnose the lesion but differentiate it from a choroidal melanoma as well. Here lies the importance of close continuous observations and serial colour fundus photographs at every visit to monitor the size and rate of growth of the tumor and to guard against malignant transformation. Such tumors hold importance from ophthalmological as well as oncological aspects. Multiple investigation modalities are needed to confirm the benign nature of the lesion though only histopathological assessment can provide confirmatory evidence.

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