

## Review Paper:

# Optic Disc Melanocytoma: A Case Report and Review



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## ABSTRACT

**Introduction:** Melanocytoma is a rare benign stationary tumor that usually appears as a pigmented lesion on the optic disk. Optic Disc Melanocytoma (ODM) can compress the optic nerve or undergo necrosis, leading to ischemic axonal loss and visual field defect, similar to those caused by glaucoma. Also, ODM often displays a clinical diagnostic dilemma due to its similarities with melanoma. Some patients have undergone enucleation because of uncertainty between both pathologies. Progressive growth and malignant transformation can be documented by close monitoring of the patient's eyes. Fundus examination and ancillary imaging procedures such as fundus photo, autofluorescence, B-scan ultrasonography, fluorescein angiography, and spectral-domain optical coherence tomography are powerful tools for ODM diagnosis and management.

**Case Presentation:** A 19-year-old female presented with a decrease in vision in the left eye for about 3 months. Her visual acuity was 20/20 and 20/80 in her right and left eyes, respectively. Funduscopic examination of the left eye showed a well-defined deeply pigmented brownish-black, dome-shaped nodular mass covered the entire optic disc with the normal-appearing overlying vitreous, macula, and surrounding retina. Short-wave autofluorescence revealed hypo-autofluorescence on the pigmented mass lesion. The patient's condition did not change significantly over 2 years of follow-up. The diagnosis was made as ODM.

**Conclusions:** Melanocytomas grow very slowly over several years or remains stable, in contrast to malignant melanoma. Although ODM tends to have benign behavior, it may adversely affect visual function. Yearly fundus examination is necessary for monitoring growth and detecting malignant transformation. Visual loss can result from optic neuropathy or retinal vascular obstruction. In suspicious cases, close follow-up with serial fundus photographs is essential, even though the malignant transformation is exceptional.

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## 1. Introduction

**O**ptic Disk Melanocytoma (ODM) is a rare, deeply pigmented benign variant of nevus that typically occurs on or adjacent to the optic disc. Clinically these patients are usually asymptomatic, and the nevus is characterized as a dark brown to the black lesion with feathery margins in the optic disk (1, 2). ODM may cause compression of the optic nerve or undergo necrosis, leading to ischemic axonal loss and visual field defects, similar to those caused by glaucoma (3-5).

Regarding the clinical presentation and vision, life-threatening melanoma represents a critical differential diagnostic challenge because of its subtle clinical similarities with ODM (6, 7). In most cases, ODM has a distinctive clinical appearance with no tendency to grow (8). However, a delicate extension may occur in 10-15% of cases during several years, and malignant transformation is considered in about 5% of cases (3, 9). Supplementary imaging procedures, such as fundus photography, short-wave autofluorescence, fluorescein angiography, and optical coherence tomography, are essential for the management and follow-up evaluations of melanocytoma (10, 11). Optical coherence tomography angiography, as a newly advanced procedure, can provide more information on optic disc perfusion than conventional angiography in ODM cases (5). As a case of ODM can imitate life-threatening melanoma and elicit a real diagnostic dilemma that bears serious clinical implications, in the current case report, we aimed to present the clinical and imaging finding in an eye with stable large unilateral ODM.

## 2. Case Presentation

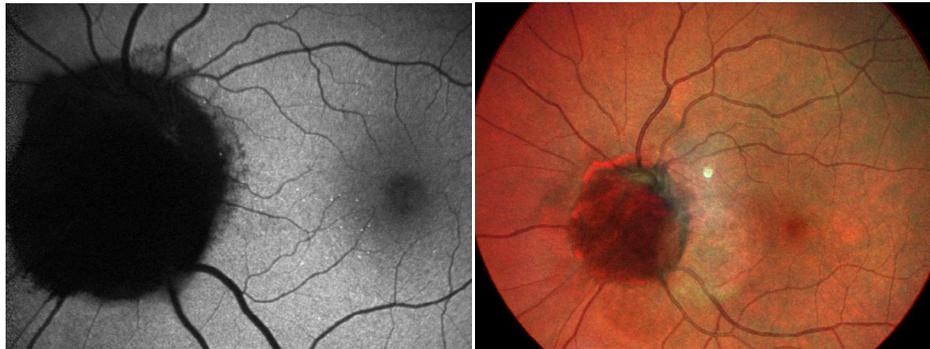
A 19-year-old Iranian female initially presented with a known decrease in vision in the left eye for about 3 months. She underwent a detailed ophthalmic examination. The best-corrected visual acuity in her right and left eyes was 20/20 and 20/80, respectively. The pupillary examination was within the normal range in both eyes. The anterior segment findings and intraocular pressures were unremarkable in both eyes. Funduscopic examination of the left eye disclosed a well-defined densely pigmented brownish-black, dome-shaped nodular mass encompassing the entire optic disc with the normal-appearing overlying vitreous, macula, and surrounding retina (Figure 1). However, the right eye ophthalmoscopy was within normal limits. Short-Wave Autofluorescence (SW-AF) using the Heidelberg Retinal

Angiography (HRA), available as the Heidelberg Spectralis, revealed hypo-autofluorescence corresponding to the pigmented masses on the optic disc. Fluorescein angiography displayed a hypofluorescent mass in both early and late phases with fine leaking from retinal telangiectasia on the tumor surface. The visual field examination exhibited blind spot enlargement. Clinically, the lesion was diagnosed as ODM. This case was followed up for over 2 years; there were no significant changes in her ocular condition.

## 3. Discussion and Review of Literature

Melanocytoma of the optic disc is a static tumor often found in an incidental routine ophthalmic evaluation. These lesions are frequently unilateral with a little preference to affect visual acuity. Central vision is normal in more than 70% of the cases (12). They have been reported to occur commonly in the sixth decade of life with a propensity for females (13). Our case demonstrates a significant clinical finding in early life with a mild decrease in visual acuity. ODM is a benign, slow-growing tumor that generally does not affect visual acuity.

Nevertheless, in the current case, compression by ODM may result in ischemic axonal loss, which may cause a clinically decrease in visual acuity. Although in the present case, visual acuity reduced recently, imaging findings failed to document critical tumor-related vision loss as mentioned above; also, over 2 years of follow-up support melanocytoma stability. ODM usually has a characteristic ophthalmoscopic feature of densely pigmented mass overlying the optic disc that may extend for a variable distance into the adjacent retina (12). The risk factors for visual loss rise if the patient experiences complications, including ischemic optic neuropathy associated with tumor necrosis, retinal vein occlusion, juxtapapillary choroidal neovascularization, or on rare occasions, malignant transformation into melanoma (12, 13). Various degrees of visual field defects can be seen in ODM consisting of an enlargement of the blind spot or substantial visual field deterioration. Also, visual field defects are present at the first visit in 80% of the cases (14). Besides, SW-AF, a noninvasive procedure according to excited endogenous lipofuscin by an exterior blue light in retinal pigment epithelium, is demonstrated to be useful for determining ODM. Guerra et al. reported that ODM becomes apparent as a prominent hypo-SW-AF lesion, and the adjacent retina was iso-autofluorescent (11). At the same time, Reznicek et al. reported that melanoma has distinct and luminous hyper-SW-AF for overlying cellular lipofuscin (15, 16). Although in most melanomas, the lesions are constant,



Journal of Pediatrics Review

**Figure 1.** Images of optic disc melanocytoma

Right: Color fundus photograph shows melanocytoma presenting as an elevated brownish-black pigmented lesion, involving most of the optic disc and adjacent retina in left eye; Left: Short-wave autofluorescence image, the melanocytoma reveals hypo-autofluorescence corresponding to the pigmented masses and adjacent retina.

**Table 1.** Reviewed literature of published cases of complicated ODM with their respective significant comments

| Authors                    | Publication Year | Study Design     | Complication (s)         | Comments  |
|----------------------------|------------------|------------------|--------------------------|---|
| Takahashi (17)             | 1979             | Two case reports |                          | Escalating pigmentation, progressive visual loss, and the leakage in the fluorescein angiogram detected in malignant transformation.  |
| Juarez et al. (18)         | 1980             | Five case report |                          | Light and electron microscopy of five melanocytomas from four patients confirmed the malignant change.  |
| Shield et al. (19)         | 1990             | Case report      |                          | Histopathology features of malignant melanoma developing in conjunction with a lesion that possessed typical clinical manifestation of an ODM.  |
| Archdale & Magnus (20)     | 1993             | Two case reports |                          | Two cases of ODM whose visual function was significantly affected by malignant transformation.  |
| De Potter et al. (6)       | 1996             | Case report      |                          | Abruptly severe visual loss after 5 years that showed malignant transformation.   |
| Meyer et al. (21)          | 1999             | Case report      | Malignant transformation | Fourfold tumor enlargement and vision loss after a 5-year period was a sign of transformation to malignant melanoma.  |
| Shields et al. (1)         | 2004             | Cases series     |                          | Although related visual loss occurred in 18% of 115 patients after 10 years and minor tumor enlargement occurred in 11% of patients after 5 years, and in 32% of patients after 10 years, the malignant transformation was documented in 2 patients (2%). |
| Shukla et al. (9)          | 2012             | Case report      |                          | Transformation of ODM into melanoma can occur over 33 years.  |
| Salinas-La Rosa (22)       | 2017             | Case report      |                          | Malignant transformation of ODM into melanoma associated with the ocular ischemic syndrome and oculocardiac reflex.   |
| Baartman et al. (23)       | 2019             | Case report      |                          | Severe vision loss raise concern for malignant transformation as it provides higher resolution optic nerve images by 7-T magnetic resonance imaging.  |
| Yamaguchi et al. (24)      | 1987             | Case report      |                          | Scattered pigment dispersion in the vitreous caused by the necrosis of the melanocytoma in an eye with ODM.   |
| Lauritzen et al. (25)      | 1990             | Case report      |                          | Tumor necrosis was the possible mechanisms responsible for seeding extracellular pigment or melanin-containing macrophages into the overlying vitreous.   |
| Shields et al. (26)        | 2001             | Case report      |                          | Abrupt visual loss in a patient with a melanocytoma does not necessarily imply malignant transformation with central retinal vascular obstruction.  |
| Font & Chaqués-Alepuz (27) | 2011             | Case report      |                          | Vitreous seeding due to tumor necrosis with the dissemination of tumor debris and melanin produced an inflammatory reaction, trabecular plugging, and ocular hypertension.  |
| Mazzuca et al. (28)        | 2012             | Case report      | Tumor necrosis           | Progressive retinal invasion and vitreous seeding from ODM.   |
| Rishi & Venkatesh (29)     | 2012             | Case report      |                          | Rapid visual loss and retinal vascular occlusion associated with ODM caused by tumor necrosis.  |
| Ackuaku-Dogbe et al. (30)  | 2013             | Case report      |                          | Report a case of necrotic melanocytoma in association with polycystic liver disease and peripheral neurofibromatosis.   |
| Guo et al. (31)            | 2014             | Case report      |                          | A 17-year-old man with ODM underwent spontaneous rupture and seeding of the vitreous with pigmented cells.  |
| Asorey-García et al. (32)  | 2015             | Case report      |                          | ODM may present with significant complications leading to significant visual loss.  |
| Agarwal et al. (33)        | 2005             | Case report      |                          | Necrotic melanocytoma of the optic disc with central retinal vascular obstruction.  |

| Authors                    | Publication Year | Study Design | Complication (s)  | Comments  |
|----------------------------|------------------|--------------|---|---|
| Gupta et al. (34)          | 1995             | Case report  | ODM progressive mass enlargement without malignant transformation | A clinically typical melanocytoma that grew progressively for 6 years without any document of malignancy.   |
| Takahashi et al. (35)      | 1984             | Case report  | Retinal arterial obstruction                                      | Central retinal artery occlusion occurred in ODM with vision deteriorated to blindness.   |
| Usui et al. (36)           | 1990             | Cases series | Ischemic optic neuropathy   | Sudden loss of visual acuity occurred, presumably due to anterior ischemic optic neuropathy induced by melanocytoma in one patient.                             |
| Tran et al. (37)           | 2006             | Case report  |   | Submacular surgery is potentially treatable for large choroidal neovascular membrane associated with ODM.   |
| Guirou et al. (38)         | 2018             | Case report  |   | Melanocytoma of the optic disc complicated by neovascularization  |
| Urrets-Zavalia et al. (39) | 2015             | Case report  | Choroidal neovascular membrane                                    | Intravitreal bevacizumab was influential in the treatment of choroidal neovascular membrane neovascularization and edema, complicating ODM.                     |
| Rouvas et al. (40)         | 2018             | Case report  |   | ODM coexisting in conjunction with polypoidal choroidal vasculopathy, promising treatment with PDT combined with intravitreal aflibercept injections.           |
| Kamisanuk et al. (41)      | 2012             | Case report  |   | Intravitreal bevacizumab can be a beneficial treatment for copy-number variation associated with ODM.   |
| Thanos et al. (42)         |                  | Case report  |   | Severe vision loss with optic disc neovascularization after hemiretinal vascular obstruction associated with ODM.   |
| García-Arumí et al. (43)   | 1994             | Case report  | ODM associated with neuroretinitis                                | Leber's neuroretinitis associated with ODM should be included in the differential diagnosis of neuroretinitis.  |
| Besada et al. (44)         | 2002             | Case report  | Compressive optic neuropathy                                      | A case report highlighting the potential impact of ODM on optic nerve head anatomy that can lead to subtle changes in the visual fields in a monocular patient. |
| Demirci et al. (45)        | 2003             | Case report  | Bilaterally in infancy  | Bilateral ODM may be associated with optic disc hypoplasia and central nervous system abnormalities such as meningioma and hypopituitarism.                     |

ODM: Optic Disc Melanocytoma; PDT: Photodynamic Therapy.

Journal of Pediatrics Review

subtle enlargement and malignant transformation have been described (15). Hence close follow-up is necessary to ascertain its benign growth.

Pathologically, ODM is a benign hamartoma, built up highly pigmented round to oval nevus cells comprising giant round cytoplasmic melanosomes with sparse cytoplasmic organelles (13). Although the tumor grows slowly with little tendency to melanoma formation, vision-threatening complications rarely occur because of its growth, compression, tumor necrosis, and malignant transformation.

According to previous reports, an increase in tumor thickness can be a marker of malignant transformation of melanocytoma, imply melanoma development after a few years of the initial appearance (1-9). Other concerns are developing vitreous seeding and vision disturbance. Although widely optic nerve tumor involvement with decreased vision can suggest malignant transformation, ischemic tumor necrosis may occur in benign melanocytoma (12). Fine enlargement has been found over the extent of several years in 10–15% of cases, but a malignant transformation is estimated to occur in 1–2% of cases (1, 43). Some

of the complicated ODM with their respective publisher findings are listed in Table 1. In conclusion, cases with ODM need yearly follow-up to detect earlier complications with photographic documentation.

## Ethical Considerations

### Compliance with ethical guidelines

Informed consent was obtained from the human subject of this study.

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### Authors contributions

All authors contributed to preparing this article.

### Conflicts of interest

The authors declared no conflict of interest.

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