

## CASE REPORT

# Central retinal artery occlusion in optic disk melanocytoma

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**Funding information**

No direct or indirect funding has been received for the presentation of the report

**Abstract**

Optic disk melanocytoma (ODM) is a rare benign tumor of the optic disk. We report a rare occurrence of profound visual loss due to central retinal artery occlusion associated with ODM in a 78-year-old female with no significant medical history. The clinical findings were supplemented by ancillary investigations.

**KEYWORDS**

central retinal artery occlusion, optic disk melanocytoma, tumor necrosis, visual loss

## 1 | INTRODUCTION

Melanocytoma, a variant of melanocytic nevus is characterized by its classic involvement of the optic disk, however sporadic involvement of the adjacent retina or choroid can also occur.<sup>1</sup> Historically, the lesion was considered primarily malignant by both clinicians and pathologists, but the benign nature of the tumor was demonstrated by Zimmerman and Garron<sup>2</sup> in 1962 and were also credited with the nomenclature. Optic disk melanocytoma (ODM) is a heavily pigmented hamartoma with a potential towards malignant transformation in 1%–2% of cases.<sup>1,3</sup>

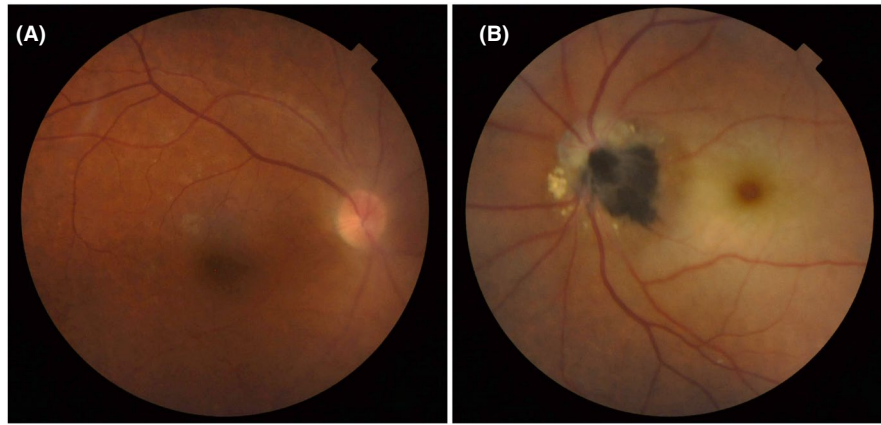
Optic disk melanocytoma is an incidental finding and hence the patients are usually asymptomatic.<sup>1</sup> Though afferent pupillary defect and visual field defects are sometimes reported,<sup>4</sup> spontaneous necrosis of the tumor or compressive optic neuropathy can result in severe visual loss in rare instances.<sup>1</sup> We report a case, which presented with severe visual loss caused by central retinal artery occlusion (CRAO) secondary to ODM.

## 2 | CASE DESCRIPTION

A 78-year-old female with no other significant medical history presented (April 2020) with sudden painless loss of vision for 5 days in her left eye (LE). On examination, the visual acuity (VA) in right eye (RE) was 6/18, which improved to 6/9 on refraction, but she denied perception of light in LE. Intraocular pressure was 13 mmHg in RE and 14 mmHg in LE, respectively. Anterior segment examination revealed relative afferent pupillary defect in LE with bilateral pseudophakia and no other significant findings. Dilated fundus examination showed early age related macular degeneration in RE (Figure 1A) and there was presence of deeply pigmented lesion with varied areas of gray to black in the region of left optic disk. The lesion covered the inferior and temporal half of the optic disk and had a fibrillar margin that extended into the adjacent nerve fiber layer of the retina. The size of the tumor was  $\sim 1 \times 2$  disk diameters, with extension into the juxtapapillary retina and choroid. The entire temporal retina within the arcades had a pale appearance except for the central

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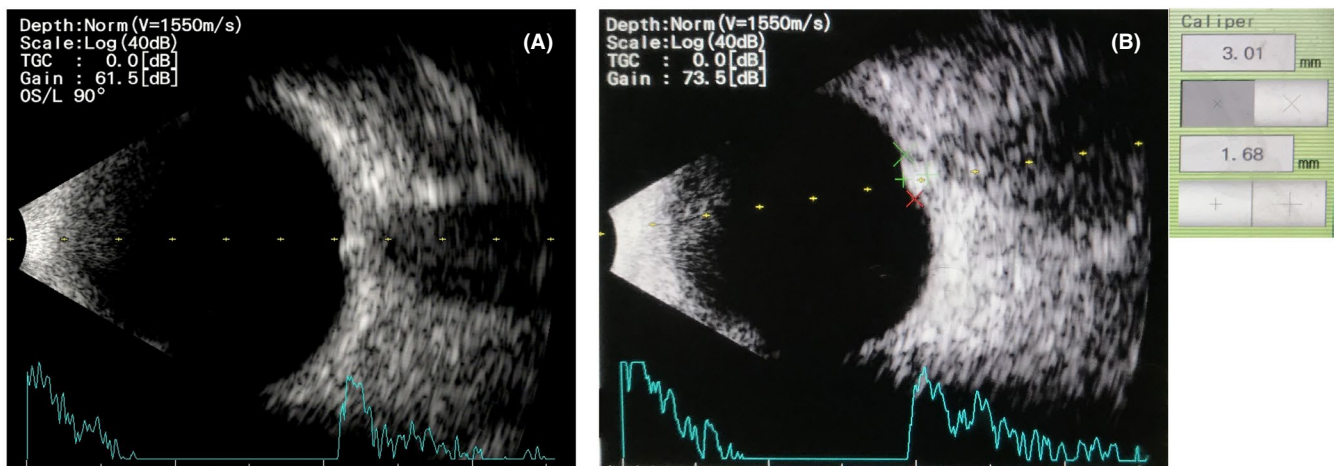
**FIGURE 1** Fundus photography at presentation. (A) Right fundus shows few age related macular degenerative lesion. (B) Left fundus with ODM that covers the inferior and temporal half of the disk. Feathery margin of the tumor is visible that extends towards the adjacent retina and choroid. Few intraretinal exudation is visible in inferior and nasal peripapillary region. Temporal retina appears edematous and pale with cherry red appearance suggestive of CRAO

cherry red appearance of the macula. The retinal arterioles were attenuated, and the retinal venules appeared dilated. Intraretinal exudation was confined to the inferior and nasal peripapillary region (Figure 1B).

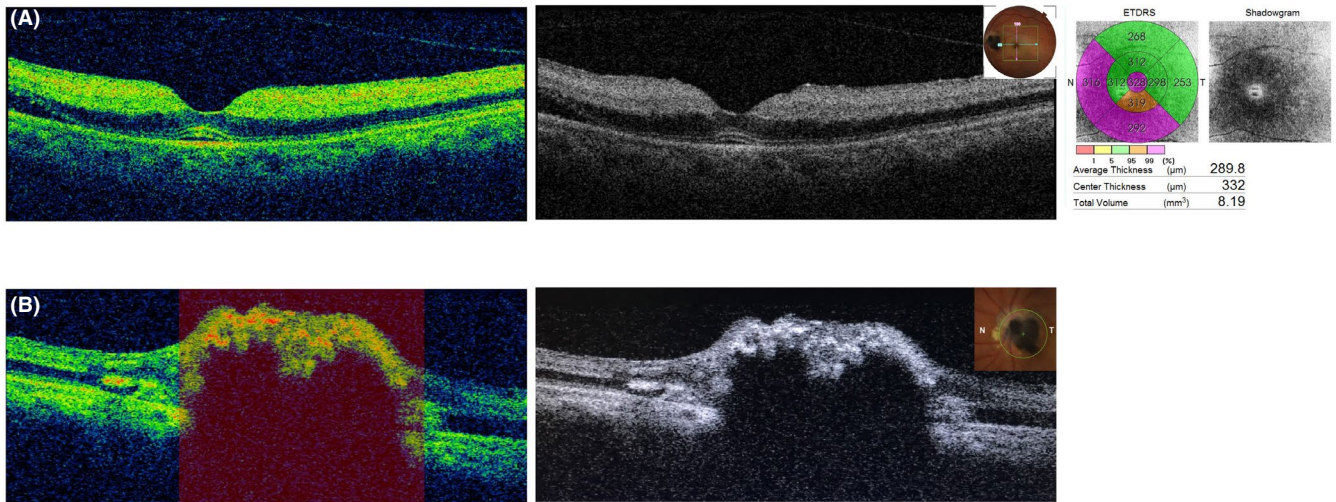
Ultrasonographic B scan showed hyperechoic signal over the disk with high surface reflectivity, moderate internal reflectivity and acoustic solidity (Figure 2A). The lesion measured 1.68 mm in thickness and 3.01 mm in its lateral extent (Figure 2). Optical coherence tomography (OCT) demonstrated retinal thickening with presence of intraretinal fluid accumulation as well as subretinal fluid and tenting of retina, nasal to the fovea in macular scan. Similarly, OCT of the disk revealed irregular hyporeflective surface contour of the tumor beneath an elevated mass of disorganized retina, which completely obscured the view of underlying tissue. The overlying retinal component had variable reflectivity (Figure 3). Fundus

fluorescein angiography (FA) revealed delayed retinal artery filling at 32 s. The tumor remained hypofluorescent throughout all the phases of angiogram. Some vessels within the mass at supero-temporal aspect appeared hyperfluorescent in the early arterial phase mimicking tumor circulation. There was presence of diffuse hyperfluorescence at the base and superior aspect of the lesion with late leakage (Figure 4).

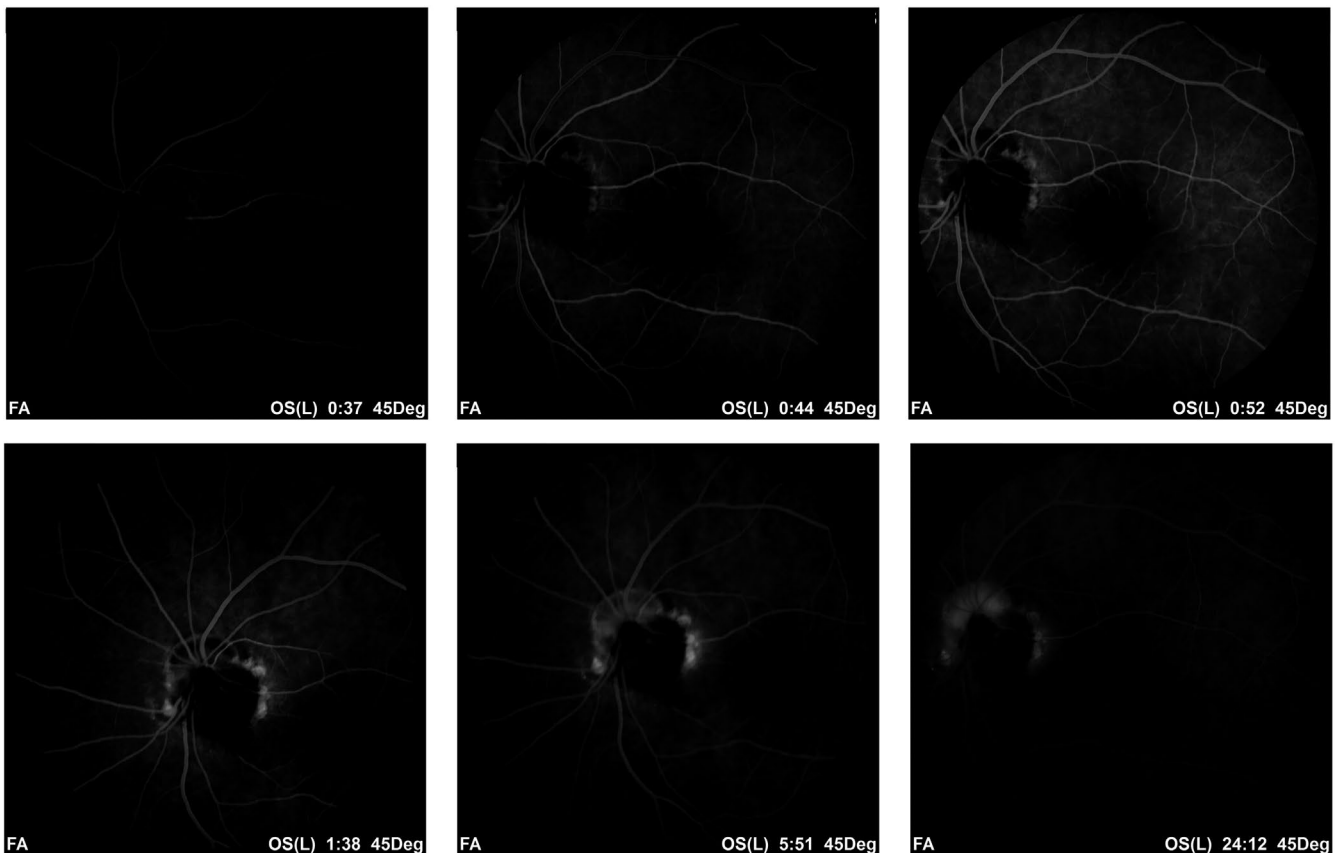
The lesion was not visible in the magnetic resonance imaging of the brain and orbit, neither optic nerve invasion nor extra-scleral invasion was evident (Figure 5A,B). However, the lesion was disclosed in the screening computed tomography scan, which showed hyperdense lesion of  $0.1 \times 0.3$  cm on the optic nerve head of LE (Figure 5C). Internist consultation was advised for systemic evaluation to rule out possible inflammatory, vascular, or neoplastic etiology including sickle cell trait as well as metastatic workup, the results of



**FIGURE 2** Ultrasonographic B scan of the LE. (A) High surface reflectivity of the tumor with acoustic solidity. Corresponding amplitude scan. (B) Measurement of the tumor dimension



**FIGURE 3** Optical coherence tomography images and corresponding grayscale image with scanned region of the fundus (inset). (A) OCT macula shows intraretinal fluid and macular edema. Retina nasal to the fovea shows tenting as well as presence of subretinal fluid. (B) OCT disk shows disorganized retinal tissue with variable intensity obscuring the details of underlying tissue. Hyperreflective dots can be appreciated within the disorganized retinal tissue overlying the tumor



**FIGURE 4** Fluorescein angiography of LE showing delayed filling of the retinal arteries and veins (timer in seconds included). The tumor overlying the disk is hypofluorescent in all phases of FA. Prominent vessel within the mass is visible in top right picture. Diffuse hyperfluorescence is present around the mass lesion during late phase

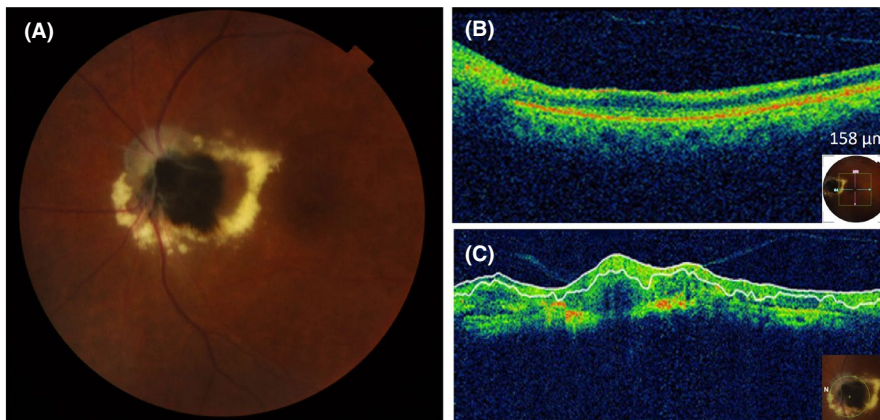
which were negative or within normal limits. On the basis of clinical examination and investigation findings, a diagnosis of LE ODM with CRAO was made. The patient was kept on

periodic follow-up and at the end of a year's review (April 2021), the size of the tumor was static, however OCT macula showed gross retinal atrophy (Figure 6).





**FIGURE 5** Neuroimaging. (A) Magnetic resonance imaging (MRI) showing orbit and brain in T1 phase. (B) MRI in T2 phase. The mass lesion is not visible in both phases of MRI. Similarly, no extra scleral or optic nerve involvement is seen. (C) Computerized tomography scan of head and orbit. The tumor is visible as hyperdense lesion just overlying the optic disk in LE (yellow arrow)



**FIGURE 6** Fundus photography and OCT at the end of 1 year. (A) The size of the tumor is stable and presence of increased exudation around the pigmented mass. (B) OCT macula shows gross retinal atrophy with 158  $\mu\text{m}$  central thickness. (C) OCT disk also reveals consistent disorganized retinal tissues

### 3 | DISCUSSION

The variants of pigmented intra-ocular lesion includes nevi, melanocytomas or melanomas that could arise from any part of the uveal tissue or retina, while melanocytes of the lamina cribrosa is the origin of pigmented lesions of the optic nerve.<sup>5</sup> Most of the melanocytomas are discovered only during routine ocular examination as they do not cause significant visual impairment.<sup>6</sup> Exudative retinal detachment involving the fovea or neuroretinitis from tumor necrosis could lead to slight deterioration of vision.<sup>7,8</sup> Whereas, profound visual loss is extremely rare event and can be attributed to central retinal vascular occlusion,<sup>9,10</sup> tumor necrosis,<sup>2,11</sup> or malignant transformation.<sup>1,11</sup>

Primary malignant tumor of the optic nerve head is limited to a handful of case reports,<sup>11,12</sup> but can be easily confused with clinical appearance similar to ODM. The lack of past medical record restricts us from the estimation of previous size of the tumor in our case. It is generally believed that the thickness of the tumor more than 1.5 mm

at presentation pose a significant risk for tumor growth.<sup>6</sup> The thickness of the lesion in our case was 1.68 mm but there was no evidence of pigmented vitreous seeds or other circumstantial evidence of malignancy,<sup>9,13</sup> though significant visual loss is found in cases of malignant transformation. CRAO along with presence of retinal extension of tumor and the presence of subretinal fluid were both present,<sup>6</sup> which can be attributed to complete loss of vision in our patient. The age could be the confounding factor for CRAO with incidental ODM in our case; however, we believe that the normal cardiac and neoplastic workup point towards the ODM responsible for causing CRAO. A report by Shields et al.<sup>10</sup> also indicated that the loss of vision in a melanocytoma need not necessarily suggest malignant transformation; instead, tumor growth and associated necrosis can lead to vascular occlusion and severe visual loss. Although exact pathogenesis of tumor necrosis is not known, it is hypothesized that the tumor causes vascular occlusion, which in turn leads to its necrosis and hypoxic retinopathy.<sup>14</sup> Teichmann and Karcioğlu<sup>15</sup> speculated that necrosis is the result of circulatory alterations in

a highly metabolic tumor. The compression of the nerve fibers and vascular occlusion is unrelated to the size of the tumor,<sup>5,16</sup> which is also analogous in our report of vascular occlusion despite small size of the tumor. Intraretinal exudation can present in upto 16% of cases<sup>1</sup> or it may be a sequelae of vascular occlusion.<sup>17</sup>

The pupillary abnormality is consistent with the findings by Osher et al.,<sup>4</sup> which can be ascribed to the axonal swelling secondary to retinal artery occlusion, disk compression, and alteration in retinal microcirculation by the tumor. The incidence of relative afferent pupillary defect has been reported in 9%–30% of patients with ODM.<sup>4,6</sup> The compressive phenomenon exerted by the ODM over the nerve fiber can lead to abnormality in pupillary reaction despite normal VA, which is one of the few conditions where this type of paradoxical pupillary reaction can occur.<sup>1</sup> This factor also explains the propensity for abnormal visual field, which can occur in up to 90% of cases.<sup>4,18</sup> OCT, a non-invasive diagnostic technique provides equal information, which corresponds with histopathological analysis.<sup>19</sup> The hyperreflective dots in OCT within the disorganized and elevated retina overlying the tumor represents the branches of the central retinal artery/vein. These dots suggest the anterior displacement of vessels and perivascular distribution of melanophages and/or tumor cells or proteins and/or lipid deposits.<sup>20</sup>

The management options for an incidentally diagnosed ODM includes fine-needle aspiration biopsy (FNAB) for cytopathological analysis, enucleation, or observation.<sup>10</sup> The option of FNAB could have been utilized as a diagnostic aid to rule out malignancy in our case. The reported sensitivity and specificity of FNAB was 84% and 98% respectively.<sup>21</sup> However, the results of cytopathological analysis of FNAB may not be always conclusive.<sup>10,17</sup> In addition, we believe that the procedure is technically demanding and relatively unsafe as the tumor in our patient was only 1.68 mm in height; hence, it was avoided. The option of enucleation was also discussed with the patient owing to no visual potential and possibility of malignant transition, but the patient declined.

There are various points that are against the de novo malignant nature of the tumor at presentation in our scenario. The profound loss of vision does not necessarily signify conversion into malignancy and is related with tumor necrosis and artery occlusion. Previous size of the tumor and its progressive growth over the course of time is unreliable due to lack of previous documentation. Absence of vitreous seeding in clinical examination and lack of extra neuronal extent of tumor in ancillary imaging modality as well as negative results of neoplastic work-up, all are in favor of benign nature of the tumor in our case. Erroneous enucleation is seldom advised with better understanding

of the nature of this type of tumor.<sup>1</sup> There are few reports available where enucleation was performed for the suspicion of malignancy in melanocytoma for profound vision loss,<sup>17</sup> retinal vascular occlusion,<sup>10</sup> and doubtful features in B scan ultrasonography.<sup>22</sup> With consideration to all these features, we decided to proceed with observation and the patient/accompanying guardian was educated about the need of periodic review.

Melanocytomas are stationary or slowly growing and locally invasive tumor.<sup>5,23</sup> Minor enlargement in the size has been reported to occur in 10%–15% of cases.<sup>1</sup> A similar incidence was observed in a series of 115 cases, where tumor enlargement occurred in 11% by 5 years, 32% by 10 years, and 38% at 20 years. Malignant transformation was recorded in only 2% of cases.<sup>6</sup> In contrast, the growth in tumor size also may not always reflect malignancy.<sup>6,23</sup> Nonetheless, an isolated case report of malignant transformation of ODM without appreciable change in size of the tumor over a period of 33 years has also been reported.<sup>24</sup>

To the best of our knowledge, there are only few reported cases of CRAO secondary to ODM.<sup>9,10,17</sup> We believe that the presentation with profound visual loss and retinal artery occlusion as in our case, may not necessarily signify malignant transformation. It may be attributed to locally invasive nature of the tumor, leading to vascular insufficiency and tumor necrosis. Erroneous decision of enucleation can be avoided in these circumstances. Though benign, the potential of malignant transformation always exist in melanocytoma. Hence, it is imperative to review these types of patients regularly and vigilance advised for any suspicious signs of malignant transformation.

## ACKNOWLEDGEMENTS

We would like to thank the patient for giving us consent to publish the case report and accompanying images.

## CONFLICT OF INTEREST

The authors report no conflict of interest to declare.

## AUTHOR CONTRIBUTIONS

Simanta Khadka (SK), Raghunandan Byanju (RNB) and Sangita Pradhan (SP) were directly involved in management of the case. SK: was involved in preparation and verification of the manuscript and analysis of the images. The final manuscript was verified and agreed upon by all the authors. All authors: made substantial contributions to conception and design, acquisition of data, or analysis and interpretation of data; took part in drafting the article or revising it critically for important intellectual content; gave final approval of the version to be published, and agree to be accountable for all aspects of the work.

## ETHICAL APPROVAL

Written and informed consent has been obtained from the patient for the publication of the case details and accompanying images. The identity of the patient has been anonymized throughout the text. Institutional approval was not required to publish case details.

## CONSENT

Published with written consent of the patient.

## DATA AVAILABILITY STATEMENT

Data sharing not applicable – no new data generated.

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**How to cite this article:** Khadka S, Byanju R, Pradhan S. Central retinal artery occlusion in optic disc melanocytoma. *Clin Case Rep.* 2021;9:e04927. <https://doi.org/10.1002/ccr3.4927>