Korean J Ophthalmol 2023;37(2):192-194
https://doi.org/10.3341/kjo.2022.0153

An Atypical Presentation of Optic Disc Melanocytoma: A Case Report

Dear Editor,

Optic disc melanocytoma is a unilateral benign tumor of the optic disc. Usually, the optic disc melanocytoma is asymptomatic and remains a stable flat dark pigmented juxtapapillary lesion throughout the patient's life, and local complications, such as choroidal neovascularization (CNV), have been reported in less than 1% of the cases [1,2]. We describe a case report of a patient with an atypical presentation of an optic disc melanocytoma associated with CNV in which differential diagnosis of a malignant lesion was crucial to avoid unnecessary diagnostic or therapeutic procedures, such as radiotherapy. Informed consent for publication of the research details and clinical images was obtained from the patient.

A healthy young man presented with progressive visual loss in the right eye (OD) over 2 months. Visual acuity (VA) was counting fingers in his OD and 20 / 20 in his left eye. Slit-lamp biomicroscopy findings and intraocular pressure were unremarkable bilaterally. Dilated fundoscopy of the OD revealed a peripapillary dark pigmented lesion surrounded by a halo of retinal atrophy, with a slightly elevated nodular portion, near the temporal border (Fig. 1A). There was also a macular edema and two subretinal hemorrhages located inferiorly to the macula. Optical coherence tomography (OCT) of the OD revealed distortion and elevation of the retinal layers towards the nasal macula, as well as an ill-defined subretinal lesion with heterogeneous internal reflectivity near the optic disc and subretinal fluid (Fig. 1B, 1C). Ultrasonography showed a peripapillary elevated hyperechogenic lesion with a largest basal diameter of 3.18 mm and a thickness of 1.85 mm associated with shallow subretinal fluid (Fig. 1D, 1E).

Received: November 25, 2022 *Final revision:* January 3, 2023
Accepted: January 31, 2023

Differential diagnosis of the peripapillary lesion included optic disc melanocytoma with secondary CNV, choroidal melanoma, choroidal nevus, congenital hypertrophy of the retinal pigmented epithelium (CHRPE), and combined hamartoma of retina and retinal pigmented epithelium (RPE). Fine-needle biopsy is seldom necessary to distinguish between benign and malignant tumors, because ultrasound features and clinical examination yield high diagnostic accuracy. The presence of symptoms, subretinal fluid and proximity to the optic nerve are all risk factors predictive of choroidal nevus growth of and transformation into a choroidal melanoma, according to the Collaborative Ocular Melanoma Study (COMS) [3]. However, ultrasound features such as thickness <2 mm and base <5 mm (also according to the COMS), high internal reflectivity, and lack of acoustic hollowness (characteristic of melanoma), along with the juxtapapillary location and patient's age, suggested a benign pigmented lesion. A combined hamartoma of the retina and RPE was excluded, because combined hamartomas are restricted to the retinal layers and combined hamartomas are usually nonpigmented lesions. On fundoscopy, the lesion could resemble peripapillary CHRPE, except for the lack of intrinsic atrophic areas, the associated CNV, and the 1.85 mm thickness measured on ultrasound, since CHRPE is almost always a flat asymptomatic lesion. Regarding treatment options, the best approach in this case was to treat the exudation from the CNV with intravitreal injection of an antivascular endothelial growth factor (anti-VEGF). VA improved to 20 / 50 following three monthly injections of ranibizumab. On OD fundoscopy, macular edema and intraretinal hemorrhages were no longer visible (Fig. 1F). Macular OCT of the OD showed almost complete restoration of normal macular architecture, with macular edema resolution in the foveal area (Fig. 1G, 1H). The patient will be kept on appropriate follow-up to prevent recurrence of macular CNV and under surveillance for malignant transformation, requiring proton beam radiation.

Optic disc melanocytoma is a variant of melanocytic nevus, a hamartoma composed of nevus cells with intense cytoplasmatic pigmentation and uniform sizes of cells and nuclei [1,2]. Although this lesion is often asymptomatic, local complications such as disc edema and retinal vascular

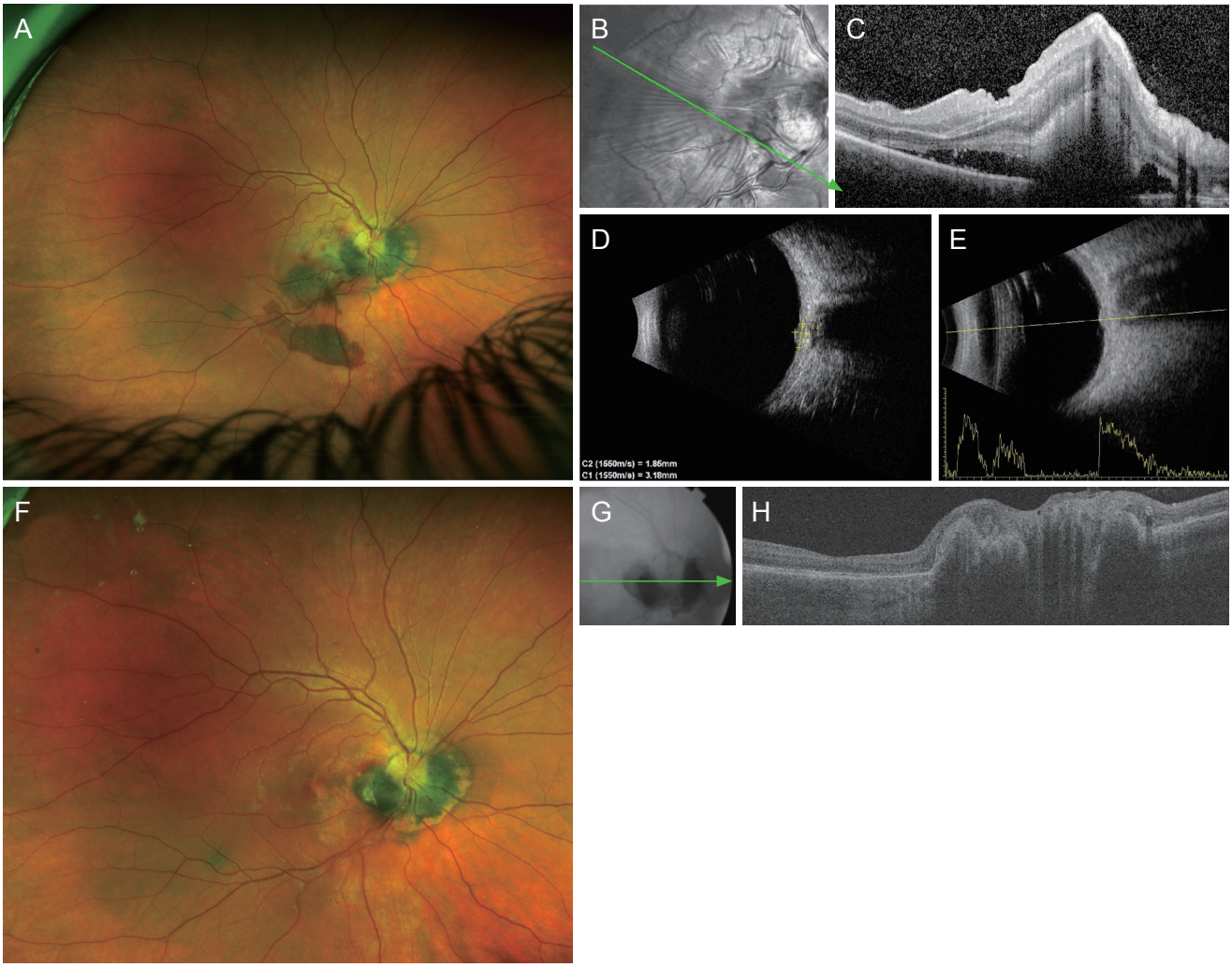

Fig. 1. (A) Ultra-widefield fundus photograph depicting a dark pigmented lesion partially encircling the optic disc, surrounded by a flat halo of retinal atrophy and with discrete elevation in the temporal side of the optic disc; there is also macular edema and two subretinal hemorrhages associated with the edema. (B,C) Optical coherence tomography B-scan (level of cross-section, arrow) showing distortion and elevation of all retinal layers with intraretinal and subretinal fluid, worsening towards the nasal side. (D,E) A-mode and B-mode ultrasound showing a peripapillary elevated hyperechogenic lesion measuring 3.18 mm of largest basal dimeter and 1.85 mm in thickness. (F) Ultra-widefield fundus photograph 1 year after treatment depicting the optic disc melanocytoma and resolution of the macular edema and subretinal hemorrhages. (G,H) Optical coherence tomography B-scan (level of cross-section, arrow) 1 year following treatment showing complete resolution of macular edema.

occlusion have been reported [1,4]. Subretinal fluid and intraretinal hemorrhages might be signs of an active CNV, which is thought to occur in less than 1% [2]. It is theorized that the tumor produces local inflammatory mediators that stimulate the formation of a CNV complex, with rupture of Bruch's membrane and ultimate bleeding and exudation [5]. Both photodynamic therapy and anti-VEGF injections are appropriate treatments for CNV [4,5]. Optic disc melanocytoma has a 1% to 2% risk of malignant transformation and patients should undergo periodic ocular examination every 1 to 2 years [1,2].

Correct diagnosis and appropriate treatment in this case were of the uttermost importance to avoid increasing patient anxiety towards his condition, and to avoid fine-needle biopsy or harmful treatments such as radiotherapy.

Conflicts of Interest: None.
Acknowledgements: None.
Funding: None.

Rosa L. Pinheiro
Department of Ophthalmology, Centro Hospitalar e Universitário de Coimbra, Coimbra, Portugal
E-mail: rosalomelinopinheiro@gmail.com

Rui B. Proença, Cristina Fonseca
Department of Ophthalmology, Centro Hospitalar e Universitário de Coimbra, Coimbra, Portugal
Faculty of Medicine, University of Coimbra, Coimbra, Portugal

References

1. Shields JA, Demirci H, Mashayekhi A, et al. Melanocytoma of the optic disk: a review. *Indian J Ophthalmol* 2019; 67:1949–58.
2. Shields JA, Demirci H, Mashayekhi A, Shields CL. Melanocytoma of optic disc in 115 cases: the 2004 Samuel Johnson Memorial Lecture, part 1. *Ophthalmology* 2004;111: 1739–46.
3. Collaborative Ocular Melanoma Study Group. Baseline echographic characteristics of tumors in eyes of patients enrolled in the Collaborative Ocular Melanoma Study: COMS report no. 29. *Ophthalmology* 2008;115:1390–7.e2.
4. Al-Halafi AM. Successful treatment of melanocytoma associated choroidal neovascular membrane with intravitreal bevacizumab. *Saudi J Ophthalmol* 2013;27:117–9.
5. Hamza HS, Moussa M, Elhusseiny AM. Management of choroidal neovascularization associated with optic disc melanocytoma with intravitreal aflibercept: a case report and review of the literature. *Case Rep Ophthalmol Med* 2019; 2019:2672798.