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Pseudo-pigmented choroidal schwannoma with extraocular extension in an elderly patient

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ABSTRACT

Purpose: To describe a rare case of clinically pigmented choroidal schwannoma with extraocular extension in an elderly patient.

Observations: We report a case of a 79-year-old Black male who presented with a clinically pigmented, juxtapapillary choroidal tumor with concern for extraocular extension on standardized ocular echography. The lesion was concerning for choroidal melanoma with extraocular extension. He had no history of cancer or known genetic disease. After an extensive discussion of management options, he underwent enucleation of the left eye. Histopathology of the tumor showed non-pigmented cells with bland, spindle-shaped nuclei and areas of Antoni A pattern along with immunostaining consistent with choroidal schwannoma with extraocular extension.

Conclusion: Choroidal schwannoma can mimic choroidal melanoma and can have a clinically pigmented appearance particularly in darkly complected individuals. This case highlights the importance of including choroidal schwannoma on the differential diagnosis for choroidal neoplasms, particularly in populations of individuals in whom melanoma is less common. Though more common in younger patients, choroidal schwannoma can present in elderly patients.

1. Introduction

Intraocular schwannomas are rare, slow-growing neoplasms that originate from sheaths of the ciliary nerves.[1,2] Tumors are typically amelanotic but can rarely be pigmented or pseudo-pigmented.[3–6] They are typically on the differential diagnosis for amelanotic choroidal lesions including amelanotic melanoma and choroidal metastasis. Distinguishing between these lesions can be clinically challenging but is extremely important given that most schwannomas are benign tumors that do not undergo malignant transformation or metastasis.[3–6]

Extraocular extension of intraocular schwannomas is also exceedingly rare with only a few reported cases in the literature.[4,6] The reason for this locally invasive behavior is not known, and the optimal management of patients with extraocular extension is unclear given the rarity of this presentation.[4,6,7] Management of choroidal schwannoma has included observation, photodynamic therapy with or without adjunct bevacizumab injection, local resection, and enucleation.[4,5,7] The majority of cases with extraocular extension have been managed with enucleation.[4]

The sex predilection of this tumor favors females (3:1) and presents at an average age of 37-years.[4] They can be sporadic or associated with systemic disorders such as neurofibromatosis type 2.[4] Here we present an extremely rare case of a clinically pigmented idiopathic choroidal schwannoma with extraocular extension in an elderly male patient illustrated with gross and histopathology images.

2. Case report

This work was reviewed by the institutional review board at the University of Iowa and was deemed non-human-subjects research. A 79-year-old Black male with past medical history of type 2 diabetes mellitus presented to the ocular oncology service for evaluation of a choroidal

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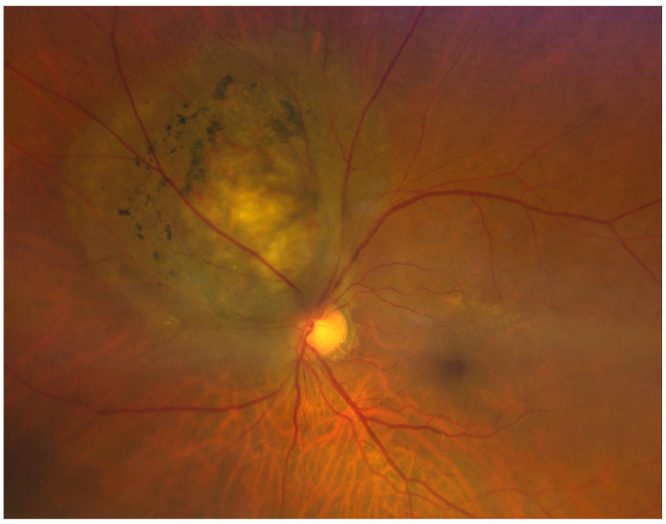

Fig. 1. Color fundus photograph of the left eye. There was a pigmented choroidal lesion abutting the optic nerve with an amelanotic central portion and overlying retinal pigment epithelium hyperplasia.

mass in the left eye that was noted on a routine diabetic eye exam. He reported that his last eye exam was four years prior to this episode without a history of a choroidal lesion. The patient was asymptomatic and had no history of cancer or neurofibromatosis. On examination his visual acuity was 20/20 in each eye without relative afferent pupillary defect or visual field deficit on confrontation. Slit lamp examination was unremarkable besides the presence of age-appropriate nuclear sclerotic cataracts.

On fundus examination of the right eye there were extramacular drusen but no diabetic retinopathy. In the left eye, there was a 12.0x11.0 × 3.9 mm pigmented choroidal lesion abutting the optic nerve from 7:00 clockwise to 1:00 with an amelanotic central portion and overlying retinal pigment epithelial hyperplasia (Fig. 1). Standardized ocular echography of the left eye showed a dome-shaped choroidal lesion with irregular internal reflectivity and no internal vascularity, measuring 3.9 mm in thickness. Posterior to the lesion, there was a 3.8 mm thick dome-shaped lesion concerning for extraocular extension, which also had no internal vascularity (Fig. 2). Optical coherence tomography (OCT) over the lesion showed a domed-shaped lesion with chronic-appearing cystoid intraretinal fluid and atrophy over the apex (Fig. 2).

The patient was counseled at length on the differential diagnosis at this point which included choroidal melanoma with extraocular extension, intra-ocular neoplasm with hemorrhage posterior to the globe mimicking extraocular extension, choroidal metastasis, inflammatory lesion, melanocytoma, and choroidal schwannoma. Management options were discussed including diagnostic biopsy, radiation (charged particle given the large area concerning for extraocular extension), observation, and enucleation. Given the concern for melanoma with extraocular extension in the context of significant barriers to transportation and ability to return for follow up care, the decision was made to proceed with enucleation of the left eye. Given the concern for melanoma, CT of the chest, abdomen, and pelvis was obtained locally without evidence of metastatic disease. He was unable to have MRI of the brain and orbits due to difficulty with insurance coverage.

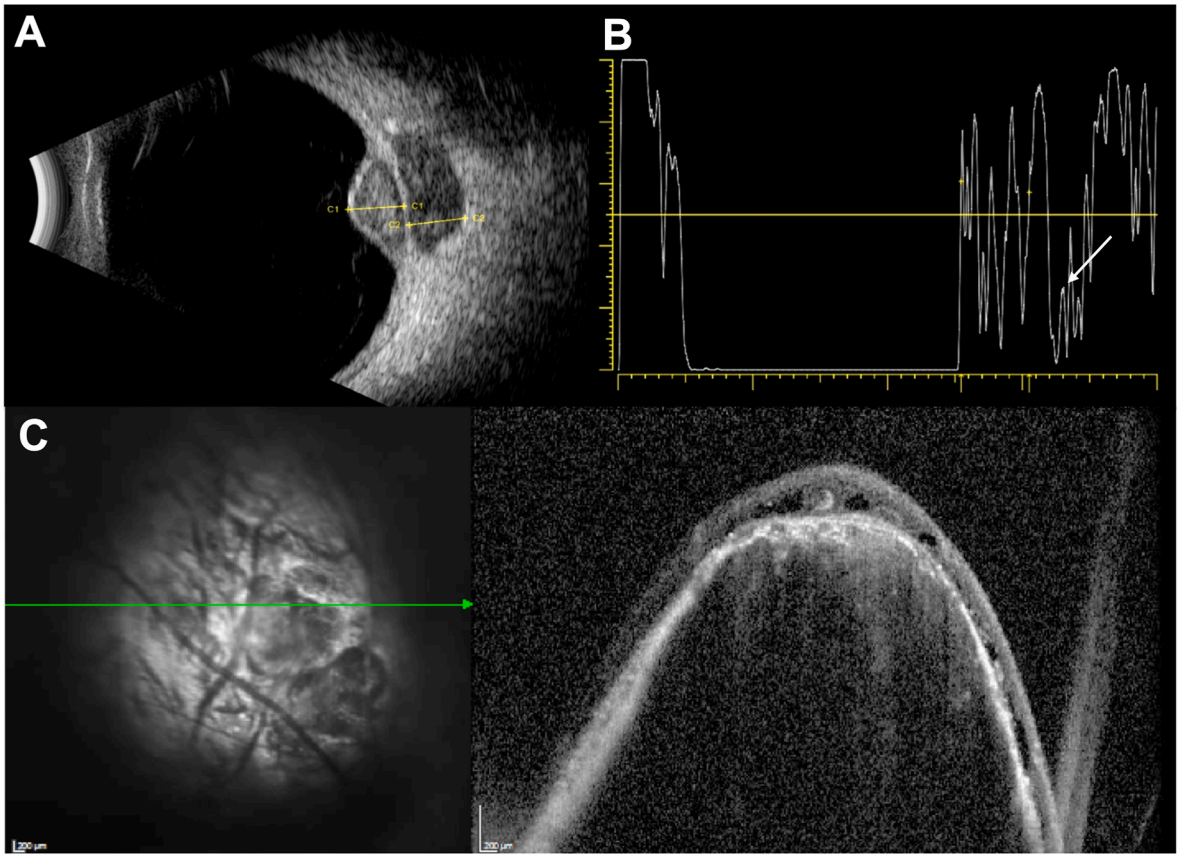

Fig. 2. Standardized ocular echography of the left eye. A) B-scan ultrasound (T11P) showed a dome-shaped choroidal lesion measuring 3.9 mm in thickness (C1) with an area of low-reflective extraocular extension posterior to the lesion (C2). B) A-scan ultrasonography of the intra-ocular lesion showed irregular internal reflectivity with very low internal reflectivity of the extraocular extension (arrow.) C) Optical coherence tomography showed chronic appearing intra-retinal fluid and atrophy of the retinal pigmented epithelium over the apex of the lesion.

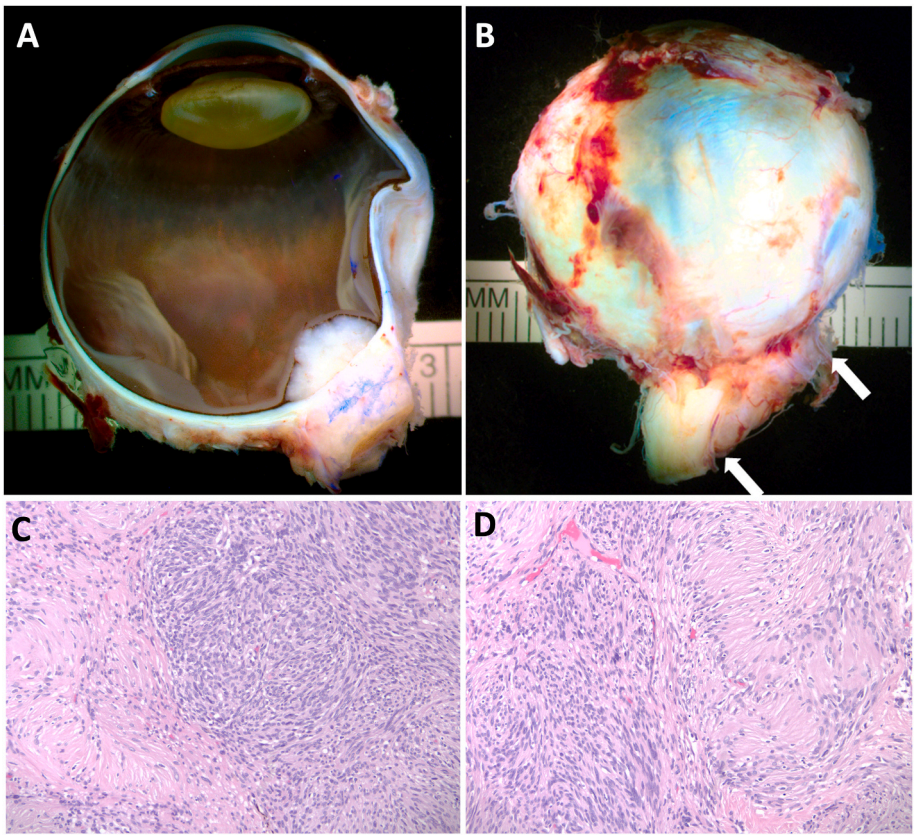

Fig. 3. Gross pathology images. A) An amelanotic dome-shaped mass was present in the posterior choroid abutting the optic disc. B) An area of off-white extra-scleral tumor adherent to the optic nerve sheath (between arrows). Haematoxylin and eosin stains (100X) showed cells with spindle-shaped nuclei in a fascicular pattern with large focal areas of palisading nuclei consistent with the Antoni A pattern of schwannoma in both the intraocular (C) and extraocular (D) tumor. No mitotic figures were present in the tumor.

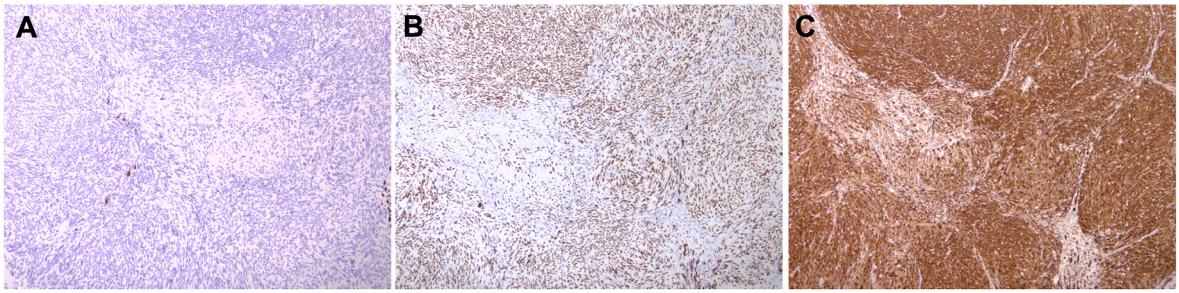

Fig. 4. Immunohistochemical staining of the tumor (50X). The lesion was Melan-A negative (A), SOX-10 positive (B), and S100 positive (C).

The patient returned for repeat ocular echography of the left eye on the day of surgery to confirm that the area concerning for extraocular extension had not decreased in size to suggest a mimic of extraocular extension such as hemorrhage. This demonstrated that the area posterior to the globe had increased in size and remained low reflective. The patient proceeded with enucleation.

On gross pathology, there was amelanotic extra-scleral tumor adherent to the superior optic nerve sheath measuring 11mm × 7.5 mm. An amelanotic dome-shaped mass was present in the posterior choroid abutting the optic disc (Fig. 3). Histopathology from both the intra-ocular and extraocular portions of the lesion revealed lesions composed of cells with slender, uniformly staining spindle-shaped nuclei arranged in fascicular pattern with scattered areas of Antoni A pattern and Verocay bodies consistent with schwannoma. Immunohistochemical staining was positive for S100 and SOX10 and negative for Melan-A and HMB45 supporting a diagnosis of choroidal schwannoma with extraocular extension (Fig. 4).

3. Discussion

Choroidal schwannoma is an extremely rare tumor that can be challenging to diagnose clinically as is highlighted by this case. Extra-ocular extension of these tumors is extremely rare and the optimal management is unclear. The low-reflective area of extraocular extension increased the suspicion for choroidal melanoma in this case. Several factors resulted in difficulty in diagnosis in this particular patient, including the fact that the patient was an elderly male (less typical for schwannoma), and that melanoma would be less likely to occur in Black patients.[4] The irregular internal reflectivity and absence of vascularity of the intra-ocular portion of this lesion were more typical of the echographic findings described in choroidal schwannoma, as opposed the low-reflectivity and vascularity typically seen in choroidal melanomas.[4] Diagnostic biopsy of the lesion prior to treatment would have been ideal, though fine needle aspiration biopsy of the intra-ocular portion of the lesion may have been less useful given that the cells of

a schwannoma can mimic amelanotic melanoma on cytology. MRI can aid in diagnosis in some cases as melanoma is often hyperintense on T1 spin-echo weighted imaging while schwannoma is often isodense to brain tissue.[4,8] Our patient had multiple barriers to care including access to transportation to our facility limiting follow up visits and his ability to undergo biopsy or have an MRI prior to treatment.

Observation of small choroidal schwannoma is reasonable given the slow growth rate and absence of metastatic potential, but larger lesions can progress to cause vision loss and most cases with extraocular extension have been managed with enucleation either due to local effects of the tumor or concern that the lesion represented melanoma.[4,5] It also must be considered that some of these lesions may begin in the sclera and spread in both directions, making identification of the primary tumor difficult to identify and resect. Tumor resection has been performed in some cases but would have been difficult in this case given the juxtapupillary location and extraocular extension.[5,7] These tumors are known to be radiation resistant limiting options for brachytherapy for globe salvage.[4,9] Given the inability to rule out melanoma, the decision for enucleation was made.

The lesion appeared clinically pigmented in this case which also complicated diagnosis. While gross pathology showed that the lesion was amelanotic, we suspect that the clinically "pseudo-pigmented" appearance was secondary to overlying retinal pigment epithelial changes as mentioned by Dong et al.[5] Schwannomas arising in areas with normal surrounding pigment can have a clinically pigmented appearance as described by Udyaver et al. in a case of a ciliary body schwannoma with episcleral extension and a pigmented appearance due to the overlying pigmented ciliary body epithelium.[6] This is an important feature to be aware of in more darkly complected individuals such as our patient and in the case reported by Shields et al.[3] Schwannoma should be on the differential diagnosis for clinically pigmented lesions particularly in populations of patients in which melanoma is less common.

4. Conclusion

Choroidal schwannomas are rare tumors that rarely have extraocular extension and can mimic choroidal melanoma. These lesions are generally amelanotic but can have a clinically pigmented appearance especially in populations of more darkly complected individuals. Schwannoma is important to keep on the differential diagnosis for choroidal lesions and management must be tailored to the individual patient and tumor characteristics.

5. Patient consent

The patient gave verbal consent to publish this case which was transcribed into the electronic medical record.

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CRediT authorship contribution statement

Arnulfo Garza Reyes: Writing – review & editing, Writing – original draft. **Chad Y. Lewis:** Writing – review & editing, Writing – original draft, Data curation. **Chau Pham:** Writing – review & editing, Data curation. **H. Culver Boldt:** Writing – review & editing. **Lola P. Lozano:** Writing – review & editing. **Nasreen A. Syed:** Writing – review & editing, Data curation. **Elaine M. Binkley:** Writing – review & editing, Writing – original draft, Investigation.

Declaration of competing interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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None.

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