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Partial spontaneous regression of choroidal melanoma: A case image with histopathology and gene expression profiling

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1. Introduction

Spontaneous regression of cancer is defined as the complete or partial disappearance of a tumor in the absence of treatment. While this phenomenon has been well-documented for cutaneous melanoma, it is quite rare for choroidal melanoma.

2. Case report

A 74-year-old man presented with vitreous hemorrhage and hand motions visual acuity in the right eye. An elevated amelanotic choroidal lesion was visible in the inferior periphery with associated exudative retinal detachment (Fig. 1a). B-scan demonstrated a choroidal lesion measuring 9.0 mm in thickness and 12.4 × 15.3 mm in basal diameter (9.0 × 12.4 × 15.3 mm) (Fig. 1b). The patient was referred to oncology to investigate potential source of metastasis. PET/CT was negative. Four weeks later, ultrasound revealed a dramatic reduction in size of the mass (3.7 × 10.0 × 11.3 mm) (Fig. 1c and d). Given the significant regression of the lesion, the patient declined further workup and elected observation. Four months after presentation, the lesion had further decreased in size (3.2 × 7.2 × 10.5 mm) and vitreous hemorrhage had resolved (Fig. 1e and f).

Upon one-year follow-up, the lesion had increased considerably in size (6.4 × 12.8 × 14.3 mm) (Fig. 1g–h) and a biopsy of the lesion was obtained. Biopsy of the mass confirmed malignant melanoma (Fig. 2a–d). Gene expression profiling revealed a Class IB, PRAME-positive tumor with mutations in *GNA11* and *SF3B1*, suggesting low-intermediate metastatic risk. The patient underwent plaque brachytherapy and transpupillary thermotherapy, resulting in tumor regression without evidence of metastases two years after treatment.

3. Discussion

The exact mechanism of spontaneous regression is unknown; it has been linked to hyperactivation of cell-mediated immunity, hormonal influences, necrosis, and vascular insufficiency.[1] To our knowledge, only eight cases have been reported, only two of which have documented histological confirmation of melanoma.[2,3] There do not appear to be clear patterns for predicting spontaneous regression, though three of the cases were associated with ocular pain and inflammation.[3] Here we provide imaging of spontaneous regression with histologic confirmation and gene expression profiling. These gene expression profiling results suggest low metastatic risk for this patient.

4. Conclusion

Spontaneous regression may rarely be observed with choroidal melanoma, which may inadvertently delay treatment. Close surveillance and a high index of suspicion are essential for diagnosis in these patients.

Patient consent

Verbal informed consent was obtained from the patient.

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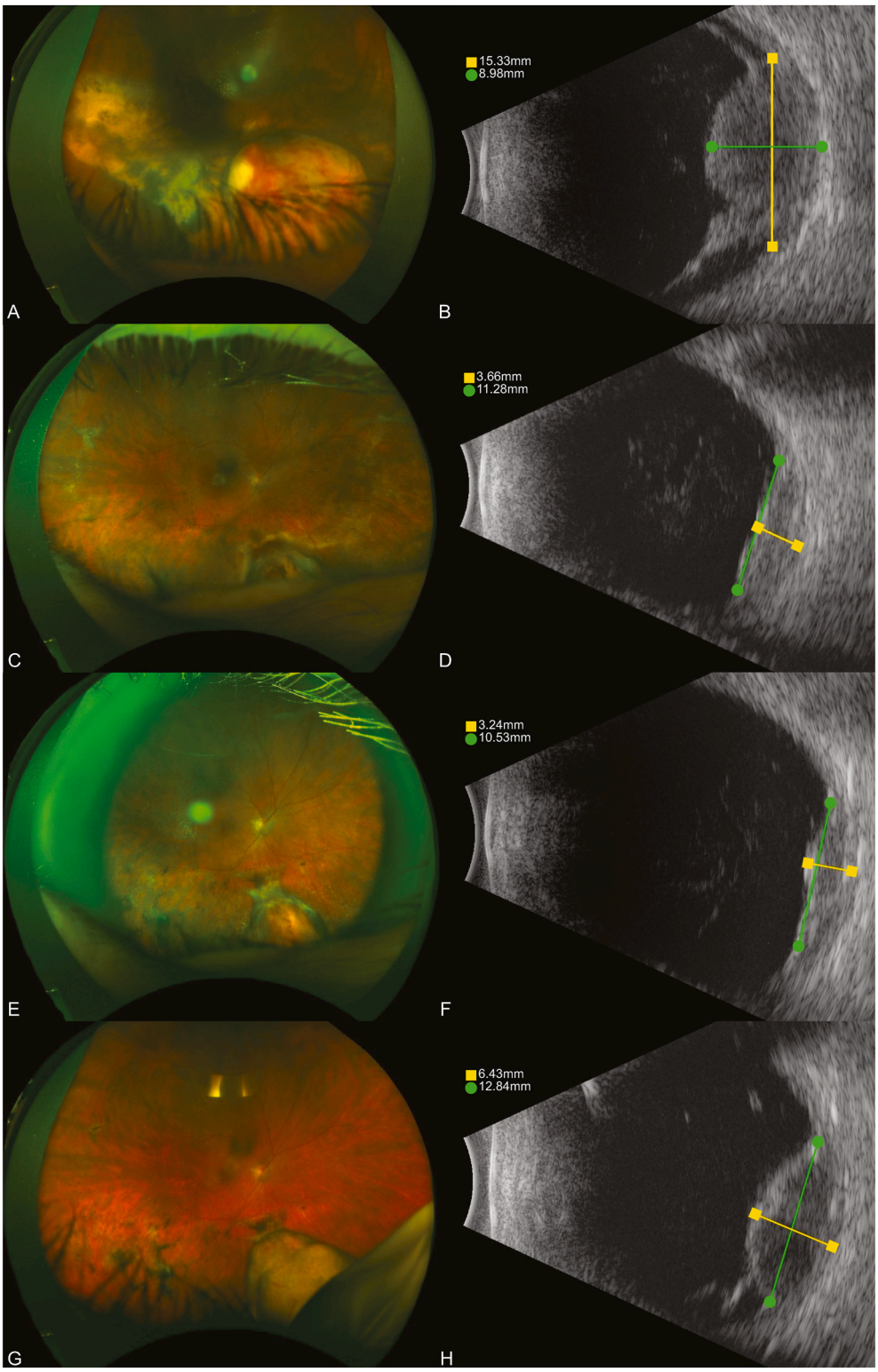

Fig. 1. On presentation, photo of elevated choroidal lesion with vitreous hemorrhage (a) and ultrasound showing dome-shaped lesion with medium internal reflectivity and associated serous retinal detachment (b). Imaging sequences demonstrating regression at four weeks (c–d) and four months (e–f) after presentation. One year later, subsequent increase in tumor dimensions and height (g–h).

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Authorship

All authors attest that they meet the current ICMJE criteria for Authorship.

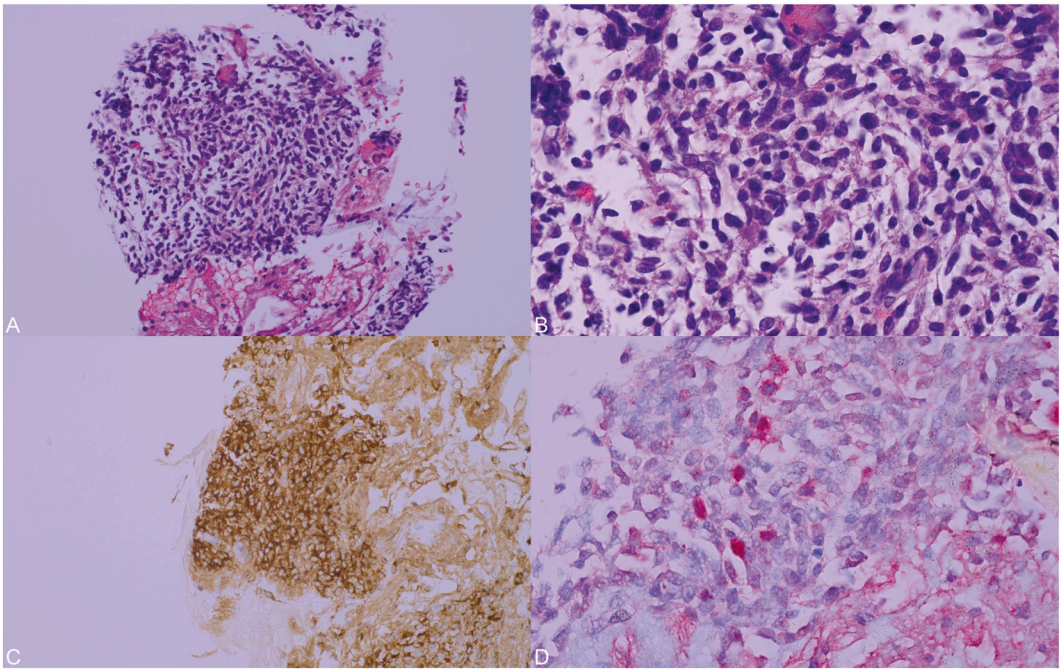

Fig. 2. Light microscopy at 400 × (a) and 1000 × (b) showing atypical spindle cells. Immunohistochemistry showing pan melanoma (c) and SOX-10 positivity (d).

Declaration of competing interest

The authors declare the following financial interests/personal relationships which may be considered as potential competing interests: Dan S. Gombos is a paid consultant for Putnam consulting, Seagen and an unpaid consultant for Castle Biosciences, Aura Biosciences. All remaining authors declare that they have no conflicts of interest.

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