Pigmented Epithelioid Melanocytoma of the Cheek, Orbit, and Intracranial Cavity: A Case Report

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Established Facts

- Pigmented epithelioid melanocytomas (PEMs) of the skin are infrequently seen in ophthalmology and are distinct entities from ocular melanocytomas.
- PEM tends to occur as a slowly growing, darkly pigmented skin nodule and has a low-grade malignant potential.

Novel Insights

- While PEM involving the conjunctiva has been reported in the literature, this is the first report of orbital involvement with satellite lesions.

Keywords

Pigmented epithelioid melanocytoma · Pathology · Pediatrics · Orbit · Surgery

Abstract

Pigmented epithelioid melanocytoma (PEM) of the skin has been rarely reported in ophthalmology. The purpose of this case report is to present a young male born with a progressive, hyperpigmented lesion involving the orbit and intracranial cavity diagnosed as PEM. The case is unique given the young age and the size, multifocality, and growth of this tumor. Identification of this lesion is paramount due to its low-grade malignant potential.

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Introduction

Pigmented epithelioid melanocytoma (PEM) of the skin has been rarely reported in ophthalmology [1]. The term “melanocytoma” itself can refer to indeterminate melanocytic lesions not belonging to any tumor classification. This can lead to confusion and miscommunication across specialties. Most ophthalmologists are familiar with the term “melanocytoma” as a benign intraocular lesion composed of highly pigmented melanocytes occurring near the optic nerve head [2]. Ocular melanocytomas are now known to occur in the choroid, ciliary body, iris, and sclera in a condition referred to as “ocular melanocytosis” [3]. In those locations, the pigmented cells are recognized to be cytologically

identical to those that were initially reported as optic disc melanocytomas and can be predisposed to transform into malignant melanomas [3, 4]. They differ from nevi in their intense pigmentation and histopathology of uniformly large, plump, jet-black, polyhedral melanocytes containing giant melanosomes and small nuclei. These melanocytomas are a different entity from PEM, which has a specific histopathologic classification discussed herein [5].

We present a young male born with a large hyperpigmented lesion involving the orbit and intracranial cavity diagnosed as PEM.

Case Report

An 11-month-old male presented for consultation with a hyperpigmented lesion of the right face present since birth and three similar lesions on his right forearm, right finger, and upper arm (Fig. 1). The patient had central, steady vision in both eyes with normal ocular motility. An initial fundus examination was normal, with cup-to-disc ratios of 0.4 bilaterally. Magnetic resonance imaging demonstrated a large mass deep within the right face centered in the region of the anterior aspect of the right infratemporal fossa, measuring roughly $3.8 \times 4.9 \times 5.2$ cm. It invaded the right orbit, causing some remodeling of the lateral bony wall and right zygoma, as well as causing the right globe to be displaced medially, with resultant kinking of the right optic nerve. The imaging characteristics of this mass showed T1 shortening, signal dropout on the T2-weighted sequence, and some minimally heterogeneous enhancement. The clinical differential diagnosis included congenital nevus or congenital melanoma.

A biopsy of the facial lesion showed a broad and thick, heavily pigmented melanocytic neoplasm throughout the dermis with very little intervening stroma and subcutaneous fat (Fig. 2, 3). The epidermis was not involved, and a grenz zone was present. There were epithelioid melanocytes with distinct nucleoli and abundant cytoplasmic melanin throughout the dermis in hexagonal arrangement. This predominant population of cells had rounded polygonal features with uniform low-grade cytomorphological features, bland-appearing nuclei, and an absence of appreciable mitotic activity. Ki-67 staining confirmed a very low mitotic count. There was a component of spindle cells present as isolated foci. Pathology from the other anatomic locations showed similar dome-shaped, heavily pigmented lesions with epithelioid melanocytes with distinct nucleoli and abundant melanin in the cytoplasm. Cellular pleomorphism, tumor necrosis, and mitoses were absent in all of these lesions.

The patient underwent surgery with ophthalmology, neurosurgery, and plastic surgery. A large portion of the zygoma, the parietal bone, and sections of the dura were resected due to infiltration. The tumor appeared to extend to the dura but not through it. The intracranial aspect of the tumor was not resected due to difficult surgical access.

Following surgery, the patient developed ptosis and amblyopia of the right eye. At 3 years, additional resection was performed to remove residual tumor. At the latest follow-up at the age 5 years, the right eye has a best corrected visual acuity of 20/125 and the tumor has not shown signs of further growth.

Fig. 1. External photograph of the orbital mass in the clinic (a) and the operating room (b). A large hyperpigmented mass measuring roughly $3.8 \times 4.9 \times 5.2$ cm invades into the right orbit.

Discussion

We described a patient with dermal PEM. PEM is a dermatopathological diagnosis described as a melanocytic tumor similar to the epithelioid blue nevus and animal-type melanoma, which had been initially identified in other mammals [5, 6]. PEM tends to occur as a slowly growing, darkly pigmented skin nodule and has a low-grade malignant potential [6]. PEM does not spread beyond lymph nodes and tends to occur in younger patients, with median onset in the third decade of life [6]. The prognosis is not well defined, though in a study of 26 patients with a median follow-up of 67 months, few adverse outcomes were observed [7].

Fig. 2. Histopathological findings. **a** Heavily pigmented, melanocytic neoplasm present throughout the dermis with little intervening stroma and subcutaneous fat. H&E. $\times 10$. **b** Epithelioid melanocytes with distinct nucleoli and abundant melanin in the cytoplasm throughout the dermis in hexagonal arrangement. H&E. $\times 40$.

PEM can comprise three cell types, i.e., large epithelioid cells, dendritic cells, or polygonal cells, with the large epithelioid cells required for diagnosis. They differ from epithelioid blue nevi, which lack large pigmented epithelioid cells [5]. PEM was established as a distinct entity when Zembowicz et al. [8] demonstrated its loss of a cyclic AMP-dependent protein kinase regulatory subunit. Similarly, PEMs associated with a familial lentiginosis and neoplasia syndrome known as Carney complex show a similar loss of expression of this regulatory subunit, providing further support that these lesions are an entity distinct from nevi [7]. Our patient showed no other stigmata of the Carney complex.

While PEM involving the conjunctiva has been reported [1], this is the first report of orbital involvement with satellite lesions. The young age of the patient, along with the size, multifocality, and growth of this lesion, makes it

Fig. 3. Bleached, stained section of epithelioid melanocytes in the hexagonal arrangement. $\times 10$.

a unique case. Proper recognition and identification of this rare entity as a benign lesion can have very important consequences for the subsequent care and management of these patients.

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Statement of Ethics

The patient's substitute decision maker provided written informed consent regarding participating in this case report, and all study protocols were conducted according to the tenets of the Declaration of Helsinki.

Disclosure Statement

The authors have no financial or proprietary interest in the materials presented herein.

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