

Case report

Pilomatrixoma in a child mimicking a ruptured epidermal cyst clinically and histopathologically: Case report

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ABSTRACT

Introduction and importance: Pilomatrixoma is a superficial benign skin tumor that originates from the matrix cells of the hair follicles. It presents more frequently during the first two decades of life and usually involves the head and neck, most often in the eyelid or eyebrow area.

Case presentation: We present a case of pilomatrixoma, which appeared at the age of 14 years with history of recurrent inflammation and discharge mimicking a ruptured epidermal cyst.

Discussion: Pilomatrixomas are often confused clinically with other benign masses, encountered in the clinical practice more frequently like dermoid cysts and epidermal inclusion cysts. The rate of accurate preoperative diagnosis ranges between 0%–30% and the correct diagnosis can be established only after excision and histopathological examination. Our case demonstrates an atypical presentation of pilomatrixoma as an epidermal inclusion cyst.

Conclusion: Ophthalmologists and ocular pathologists should be aware of the atypical presentation of pilomatrixomas to ensure early accurate diagnosis and curative treatment.

1. Introduction

Pilomatrixoma, also known as calcifying epithelioma of Malherbe, is a benign skin neoplasm that arises from hair follicle matrix cells. It is common among the pediatric population with peak presentation within the first two decades of life [1]. The periorbital area is considered one of the most common locations of pilomatrixoma in the head, with the upper eyelid and brow being the primary locations [1–3]. Pilomatrixoma is often misdiagnosed clinically, and the correct diagnosis can only be established after surgical excision and histopathological examination [4]. The presence of two cell populations, the anucleated shadow cells and the basaloid cells, which lack nuclear features of malignancy, is specific for pilomatrixoma [1].

Reports of pilomatrixoma are sparse in the literature, however they mostly stress on the low rate of clinical suspicion and accurate preoperative diagnosis. This article reports a case of pilomatrixoma arising from the upper eyelid in a 14-year-old girl with an atypical

misleading clinical presentation and unique corresponding histopathological appearance. Similar presentation to our case has been described in only 2 previous reports that will be discussed further in comparison to ours.

This case report was prepared in accordance with the ethical standards and the Helsinki Declaration. No trial of new drugs or therapy is applicable in this case. Case reports do not require Ethical approval in our institution. However, a general written informed consent was taken from the guardian of the patient, which includes permission for anonymous use of information and photos for reporting. This case report has been prepared in line with the updated SCARE 2020 criteria [5].

2. The case

A healthy 14-year-old female presented to our clinic with a right upper eyelid painless mass that was stable in size over a period of six months. The patient reported an episode of pain and redness with some

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sticky material coming out from the lesion. She was treated outside with topical and oral antibiotics with an apparent resolution of the symptoms; however, the swelling persisted. There was no significant past medical or surgical history. Her family history was negative as well with no features or history suggestive of genetic abnormalities.

On examination, the lesion was located below the lateral aspect of the eyebrow. It was nodular, red in color, round, firm in consistency, and adherent to the skin but not fixed to the underlying tissue with defined margins, measuring 2 × 2 mm. The overlying and surrounding skin appeared normal, and no punctum was visible (Fig. 1). The rest of the ophthalmic examination was unremarkable.

Based on the site, the lesion's nodular nature, and the history of inflammation and discharge, a provisional diagnosis of the ruptured epidermal cyst was suspected. The diagnosis was explained to the patient and parents, and the guardians happily consented to the surgical removal of her eyelid lesion for both diagnostic and cosmetic purposes by an experienced oculoplastic surgeon. The patient underwent a successful excisional biopsy under local anesthesia and was well tolerated by the patient with no complications. Post-operatively, the patient was discharged on topical antibiotics with a 2-weeks follow-up, and the specimen was sent for histopathology.

Grossly, the specimen was a tan-colored round soft tissue lesion measuring 2 mm in diameter. Histopathologically, the hematoxylin and eosin-stained sections revealed a tumor composed of an epithelial component exhibiting the typical population of basaloid cells and other areas of eosinophilic ghost cells devoid of any nuclei. A focal area of squamous proliferating cells was seen with adjacent keratin material confirmed by reactivity to cytokeratin marker (Fig. 2A–C). Also, a mixed inflammatory cell infiltrates with multinucleated giant cells, areas of calcification were present (Fig. 2D). Based on these histopathological findings, the lesion was diagnosed as pilomatrixoma with an area mimicking an incomplete or ruptured epidermal cyst.

The patient was seen in her 2-weeks follow-up appointment with complete wound healing at the excision site and excellent cosmetic result. She was informed about the benign nature of the excised skin lesion and was expected to be discharged from our ophthalmic care after her next visit.

3. Discussion

Pilomatrixoma is a benign, slowly growing cutaneous neoplasm that arises from the matrix of the hair follicles. Up to 55% of pilomatrixoma presents in the first two decades of life, with females being more commonly affected than males with a female-to-male ratio of 1.15–1

[1]. It is found mainly in the head and neck region (64% of cases) with the periorbital area, namely, the upper eyelid and eyebrow being most frequently involved [1,2]. Other common locations include the upper extremities, the trunk, and the lower extremities [1]. Clinically, these neoplasms commonly present as an asymptomatic, firm, slowly growing, mobile nodule with a normal or hyperemic overlying skin. Sometimes, the lesions show inflammatory signs [1,6]. Helpful pathognomonic diagnostic signs are “teeter-totter” and “tent sign” [1,4]. Both can be demonstrated during clinical examination by stretching the skin over the tumor, which will show multiple facets over the lesion (tents sign), and the teeter-totter sign which can be illustrated by pressing on one edge of the lesion, this will cause the opposite edge to protrude from the skin.

Despite the well-described features mentioned above, pilomatrixoma is often misdiagnosed clinically, and the correct diagnosis can be established only after excision and histopathological examination. A literature review demonstrated that the accuracy rate of the preoperative diagnosis of pilomatrixoma ranges from 0% to 30% of cases [6]. Zloto described the largest series of periocular pilomatrixoma and confirmed the tendency of the lesion to develop in the first 2 decades of life (in 69%), the female predominance, and the accurate clinical diagnosis in only 18.75% of the cases [7]. Pilomatrixomas are often confused with other benign masses, encountered in the clinical practice more frequently like dermoid cysts, foreign body reaction, lipoma, and epidermal inclusion cysts, like our report [6]. However, this largely depends on the location of the lesion and the age at presentation. Nigro for example presented pilomatrixoma as a midline facial lesion over the bridge of the nose of an 11-month-old infant with the differential clinical diagnosis of a dermoid cyst, glioma, encephalocele, and vascular anomaly [8]. Pre-operative imaging using computerized tomography (CT) scan, magnetic resonance imaging (MRI), and ultrasonography has also limited additional value in the diagnosis of pilomatrixoma [8]. Park reported the youngest case of periocular pilomatrixoma involving the upper eyelid in a 10-month-old baby with the onset at the age of 3 months and stressed on the difficulty of diagnosing such cases clinically especially that fine needle aspiration might not be feasible in infancy [9]. Despite the use of MRI in their case, the clinical diagnosis was only reached after complete excision of the lesion. The pre-operative diagnosis in their case based on imaging was ossifying hematoma [9]. We performed careful literature search and found only 2 previously published cases of pilomatrixoma presenting as an epidermal inclusion cyst similar to our patient [4,10]. In both studies, the diagnosis of pilomatrixoma was only established after surgical excision of the lesion and histopathological examination, which highlights the significance of this report to promote recognition of this atypical presentation among ophthalmologists and emphasize the role of a thorough clinical examination, which can provide a definitive diagnosis for early, asymptomatic, and clinically unsuspected cases of pilomatrixoma. The typical histopathological features are large masses of two major cell types: basophilic cells and eosinophilic shadow cells. Inflammation, foreign body-type giant cells, calcification, and ossification can be seen as well [1,6].

Identification of this lesion is important because, although extremely rare, pilomatrixoma can undergo malignant transformation into a pilomatrix carcinoma with unusual features [6,11]. Complete surgical excision of this tumor with clear margins is sufficient and curative treatment, with excellent postoperative prognosis for cosmeses and also to prevent the risk of recurrence and the possibility of malignant transformation [1,9].

In conclusion, we are presenting an atypical case of pilomatrixoma that was clinically diagnosed as a ruptured epidermal cyst. The main aim of this report is to promote awareness of the variable wide atypical presentation of those lesions and the importance of careful clinical screening and high level of suspicion. An early, definitive, pre-operative diagnosis ensures a curative treatment by complete surgical excision and, although rare, prevents possible recurrence and malignant

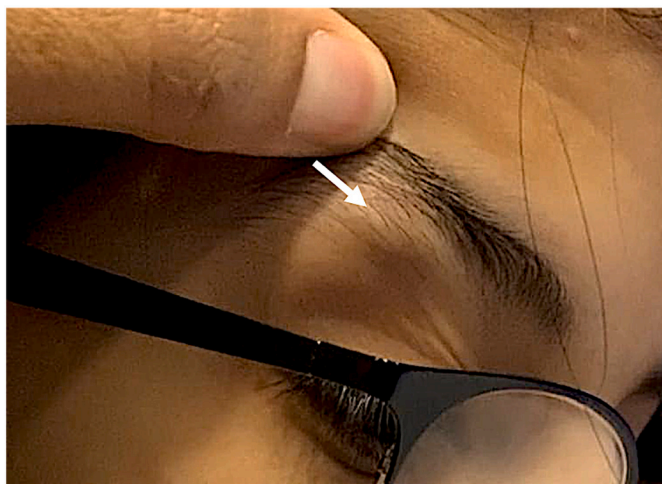


Fig. 1. Clinical photo of the right upper eyelid nodule just beneath the eyebrow (white arrow).

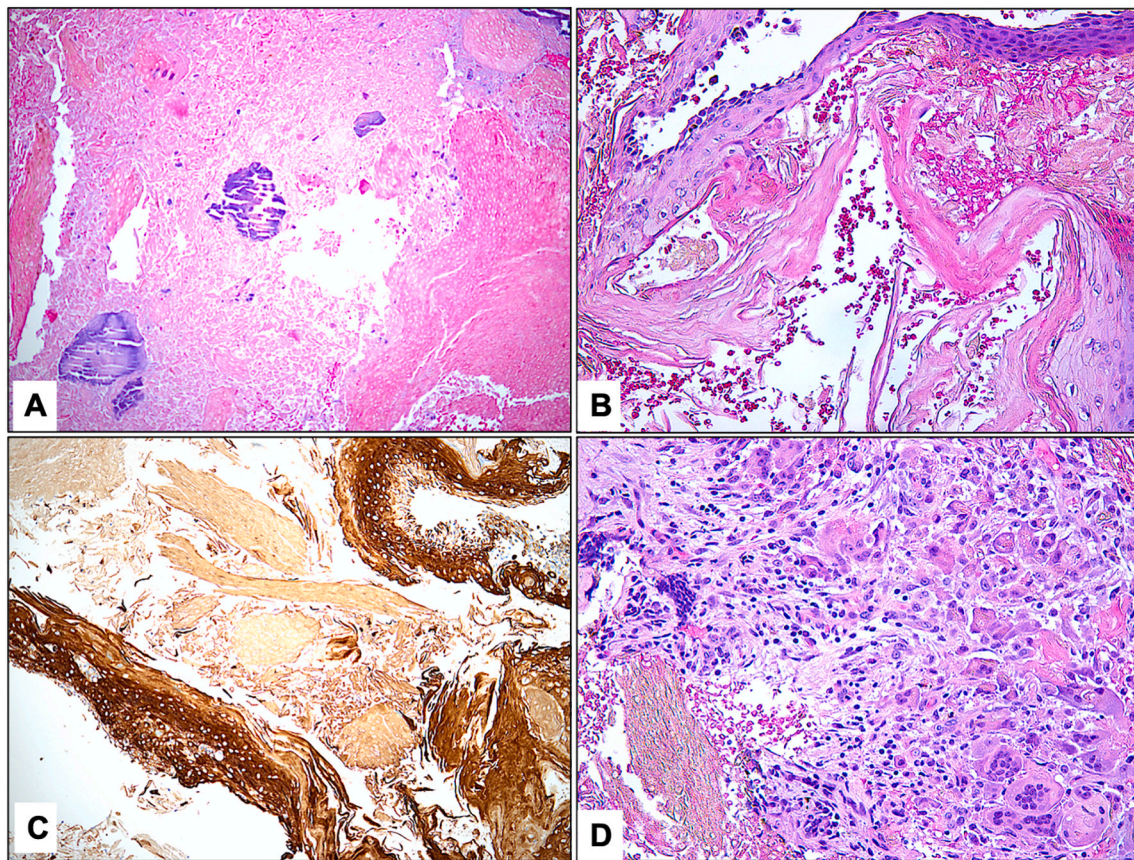


Fig. 2. A: The histopathology photo of the lesion consisting of the typical basaloid cells and the eosinophilic ghost cells in addition to calcifications (Original magnification x100 Hematoxylin and eosin). B & C: Focal area of squamous proliferating cells was seen with adjacent keratin material mimicking a wall of ruptured epidermal cyst with the squamous component expressing positive reaction to Pan-cytokeratin marker in C (Original magnification x100 Hematoxylin and eosin in B and Cytok in C). D: Higher power of the area of granulomatous reaction and foreign body-type giant cells typically seen in pilomatrixoma (Original magnification x200 Hematoxylin and eosin).

transformation.

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None.

Ethical approval

Case reports do not require ethical approval in our institution. However, information was obtained and reported in a manner that was compliant with the standards set forth by the Health Insurance Portability and Accountability Act, and the Declaration of Helsinki as amended in 2013.

Consent to participate

A written informed consent was obtained from the guardian of the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Research registration

Registration is not required for Case reports.

Guarantor

Dr. Hind M. Alkatan, MD.

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CRediT authorship contribution statement

HMA: Study design, histopathological examination, final tissue diagnosis, and overall review for editing of the manuscript as the corresponding author. WA: Chart review, data collection, literature review and first draft of the case report. OA: Literature review and first draft of the case report. DA: Treating Oculoplastic Surgeon and clinical images.

Declaration of competing interest

The authors have no financial or conflict of interest related to this work. An informed General Consent has been taken, which includes using patient's anonymous information.

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