Eyelid pilomatrixoma: morphological features, clinical rarity, and diagnostic pitfalls in ophthalmologic practice

Pilomatrixoma palpebral: características morfológicas, raridade clínica e armadilhas diagnósticas na prática oftalmológica

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ABSTRACT

Pilomatrixoma is a rare benign neoplasm derived from hair matrix cells, with a preference for the head and neck region, especially in children and adolescents. Its occurrence in adults, particularly in the eyelid, is uncommon and often leads to misdiagnosis. Clinically, it presents as a firm, painless, subcutaneous nodule with progressive growth. Definitive diagnosis is established through histopathological examination, and the standard treatment is complete surgical excision, with low recurrence rates. This article describes a case of eyelid pilomatrixoma in a young adult, highlighting its morphological features, clinical rarity, and diagnostic challenges in ophthalmologic practice.

RESUMO

O pilomatrixoma é uma neoplasia benigna rara derivada das células da matriz capilar, com predileção pela região da cabeça e pescoço, especialmente em crianças e adolescentes. Sua ocorrência em adultos, particularmente na pálpebra, é incomum e frequentemente leva a erros diagnósticos. Clinicamente, apresenta-se como um nódulo subcutâneo firme e indolor, de crescimento progressivo. O diagnóstico definitivo é estabelecido por meio do exame histopatológico, e o tratamento padrão é a excisão cirúrgica completa, com baixas taxas de recorrência. Este artigo descreve um caso de pilomatrixoma palpebral em um adulto jovem, destacando suas características morfológicas, a raridade clínica e os desafios diagnósticos na prática oftalmológica.

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INTRODUCTION

Pilomatrixoma, also known as calcifying epithelioma of Malherbe, is a benign cutaneous adnexal tumor originating from hair matrix cells.[1,2] It accounts for less than 1% of excised skin lesions and is more common during the first two decades of life, with a preference for the head and neck region.[3] Approximately 40% to 60% of cases occur in the head and neck, while eyelid involvement accounts for less than 5%.[3-5] There is a slight female predominance and a second incidence peak between 50 and 65 years of age, making its occurrence in young adults an additional peculiarity.[4]

Although typically isolated, pilomatrixoma may be associated with genetic syndromes such as Gardner syndrome, Rubinstein-Taybi syndrome, and myotonic dystrophy type 1, especially in cases with multiple lesions.[5-7] These associations underscore the importance of systemic investigation in atypical or recurrent cases.

Its clinical presentation is often confused with more common benign lesions, such as chalazion, sebaceous cysts, and lipomas, hindering early diagnosis. It usually manifests itself as a firm, painless, slow-growing subcutaneous nodule, which may occasionally show inflammatory signs or calcified areas detectable on palpation. Diagnosis is confirmed by histopathology, which reveals peripheral basaloid cells, central anucleate ghost cells, and areas of dystrophic calcification.[2,8]

The treatment of choice is complete surgical excision with clear margins, resulting in low recurrence rates when properly performed.[9] This study aimed to report a rare case of eyelid pilomatrixoma in a young adult, discussing its morphological aspects and the main diagnostic challenges in ophthalmologic practice.

This case report was conducted in accordance with the ethical standards of the institutional and national research committees and with the 1964 Helsinki Declaration and its later amendments. The study was approved by the Research Ethics Committee of the University Hospital Júlio Müller of the *Universidade Federal de Mato Grosso* (UFMT) under protocol number 7.608.588 and CAAE: 88546725.7.0000.5541. The patient signed an Informed Consent Form authorizing the publication of clinical data and images for scientific purposes.

CASE REPORT

A 39-year-old previously healthy male presented to the ophthalmology clinic with a one-month history of a nodular lesion on the left upper eyelid. The patient reported progressive growth without pain, discharge, or local inflammatory signs (Figures 1 and 2). He denied any prior trauma, surgery, or relevant dermatological history. Ophthalmological examination revealed a best-corrected visual acuity of 20/20 in both eyes. On inspection, there was a subcutaneous nodule on the left upper eyelid, approximately 1 cm in diameter, with regular borders, smooth surface, skin-colored appearance, and no pigmentary or ulcerative changes. On palpation, the lesion was firm, mobile over deep planes, painless, and without inflammatory signs. Ocular motility, slit-lamp examination, and fundus evaluation were unremarkable.

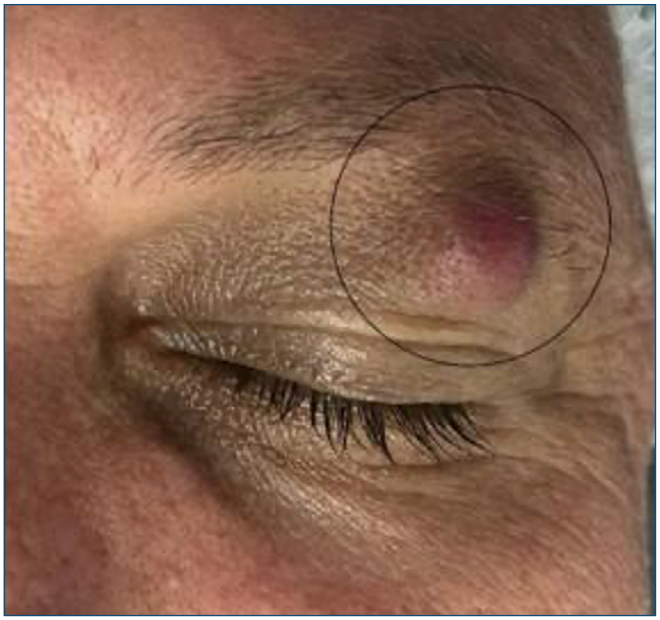

Figure 1. Preoperative view showing a nodular tumoral lesion on the left upper eyelid.

Differential diagnoses included epidermoid cyst, atypical chalazion, and benign cutaneous gland tumor, among others. The patient underwent complete excision of the lesion under local anesthesia, with a safety margin of approximately 1 mm. The specimen was fixed in 10% formalin and sent for histopathological analysis. Macroscopically, it appeared as a firm, encapsulated nodule with calcified areas visible on sectioning. Histology showed a well-circumscribed tumor composed of epithelial islands with peripheral basaloid cells and extensive central areas of keratinization with anucleate ghost cells (Figure 3 and 4). A granulomatous inflammatory reaction with multinucleated foreign body giant cells and foci of calcification were also observed. These findings confirmed the diagnosis of pilomatrixoma.[2,5]

The postoperative course was uneventful, with mild eyelid edema and no signs of infection, dehiscence, or hematoma (Figures 5 and 6). Sutures were removed on day

Figure 2. Preoperative view of the nodular tumoral lesion on the left upper eyelid.

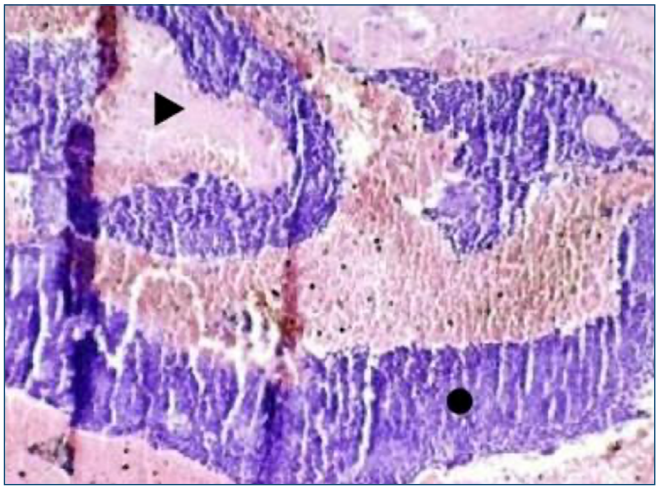

Figure 3. Histopathological appearance after biopsy of the nodular lesion, showing ghost cells with loss of nuclear basophilia.

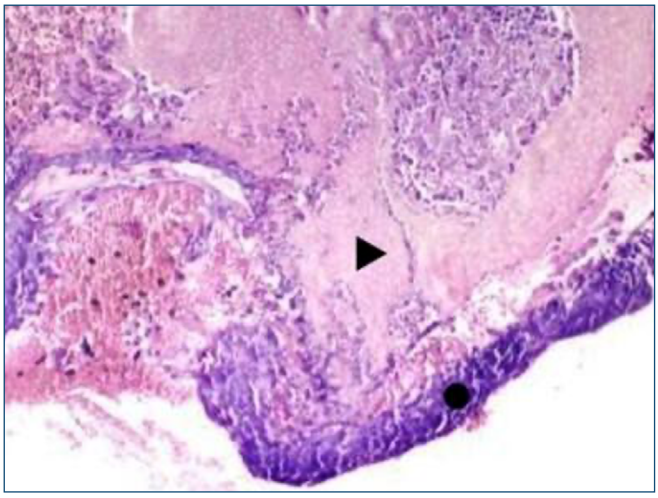

Figure 4. Histopathological appearance after biopsy of the nodular lesion, showing viable basaloid cells, similar to those of the hair follicle matrix.

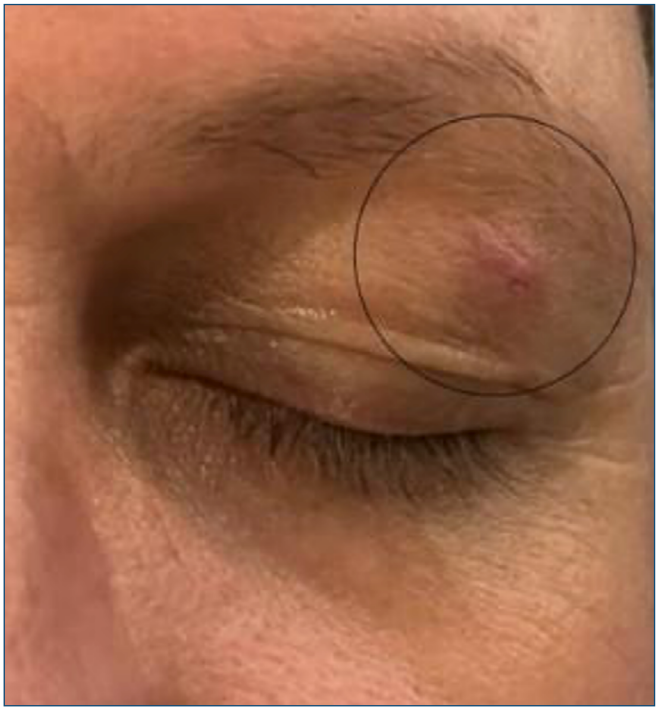

Figure 5. Postoperative aspect after excision of the nodular lesion on the left upper eyelid with eyes closed.

7. At 1-month follow-up, the scar was subtle and well-positioned along the upper eyelid crease. Clinical follow-up continued for six months with no evidence of recurrence or local complications.

DISCUSSION

The rarity of eyelid pilomatrixoma in adults makes clinical recognition particularly challenging. Studies demonstrate that, although pilomatrixoma is relatively common in the pediatric population, especially during the first two decades of life, its presentation in individuals over 30 years old is notably uncommon, accounting for less than 5% of reported cases in the literature.[2-5] This significantly low incidence in adults contributes to its under-recognition by clinicians, who may be less familiar with this entity outside the typical age group.

Figure 6. Postoperative aspect after excision of the nodular lesion on the left upper eyelid with eyes open.

Moreover, the clinical presentation of pilomatrixoma in adults is often subtle and nonspecific, typically manifesting as a firm, painless, slowly growing subcutaneous nodule without distinctive features that could easily differentiate it from more frequent eyelid lesions such as chalazion, epidermoid cysts, or benign adnexal tumors. This clinical ambiguity further complicates timely and accurate diagnosis. In many cases, pilomatrixoma is initially misdiagnosed or overlooked, leading to delayed treatment and potential enlargement of the lesion, which might increase surgical complexity and impact cosmetic outcomes.[2,8]

Additionally, adult patients may present with atypical features, such as a more rapid growth rate or associated mild inflammation, which can be misleading and erroneously suggest malignancy or infectious processes. The rarity of pilomatrixoma in this age group, combined with such nonspecific manifestations, underscores the importance of maintaining a high index of suspicion among ophthalmologists and other clinicians managing eyelid lesions. Awareness of pilomatrixoma's clinical and epidemiological characteristics is essential for early consideration in differential diagnoses, especially for eyelid nodules that do not respond to conventional treatments.[8,9] Since the eyelid is a delicate anatomical region with significant functional and aesthetic importance, misdiagnosis can lead to inappropriate interventions or repeated unsuccessful treatments, increasing patient morbidity.

Therefore, comprehensive clinical assessment complemented by appropriate imaging modalities and a low threshold for biopsy or complete excision is advisable in suspicious cases. Such a proactive diagnostic approach can improve recognition rates, enable histopathological confirmation, and ensure timely surgical management, ultimately enhancing patient outcomes and minimizing the risk of recurrence or complications.[9]

In the periocular region, pilomatrixoma is often clinically indistinguishable from other common eyelid lesions, which can lead to diagnostic challenges. It may be mistaken for chalazion, sebaceous cysts, Meibomian gland tumors, xanthelasma, dermoid cysts, or even malignant tumors such as basal cell carcinoma or sebaceous carcinoma.[2,8] This broad differential diagnosis is due to overlapping clinical features such as nodularity, firmness, and slow growth. However, pilomatrixoma may occasionally exhibit a more rapid enlargement or subtle changes in consistency that are not typical for benign lesions, further complicating clinical assessment.[10]

Given this, it is crucial for clinicians to maintain a high index of suspicion when evaluating firm, painless eyelid nodules, particularly those that demonstrate progressive growth. This vigilance should be upheld even in the absence of classic inflammatory signs like redness or tenderness, or pigmentary alterations that might suggest malignancy. Early consideration of pilomatrixoma in the differential diagnosis can prompt timely biopsy or excision, avoiding mismanagement or delay in definitive treatment. Ultimately, thorough clinical evaluation combined with awareness of this entity is vital to distinguish pilomatrixoma from other eyelid pathologies and guide appropriate patient care.

High-frequency ultrasound is a valuable, non-invasive imaging tool that can aid in the preoperative assessment of eyelid lesions suspected to be pilomatrixomas. On high-frequency ultrasound, pilomatrixoma typically presents as a well-defined, hypoechoic mass characterized by internal hyperechoic foci, which corresponds to areas of calcification within the lesion. These calcifications often produce posterior acoustic shadowing, a feature that helps to distinguish pilomatrixoma from other soft tissue masses. The clear margins observed on ultrasound imaging also assist in surgical planning by delineating the extent of the tumor.[9] However, despite these useful imaging characteristics, high-frequency ultrasound cannot

definitively establish the diagnosis, and its findings must be interpreted within the broader clinical context.

The definitive diagnosis of pilomatrixoma relies on histopathological examination, which reveals highly characteristic features that experienced pathologists can readily identify. Microscopically, the tumor is composed of epithelial islands with uniform basaloid cells at the periphery. Centrally, there are large areas filled with anucleate "ghost" cells, which exhibit eosinophilic cytoplasm and a loss of nuclear staining (basophilia), reflecting keratinization.[1,2,5]

Additionally, dystrophic calcification is commonly present within the lesion, often accompanied by a granulomatous inflammatory response containing multinucleated giant cells. In rare cases, metaplastic ossification may also be observed, further highlighting the diverse histological patterns of this neoplasm. These distinctive histological hallmarks confirm the diagnosis and differentiate pilomatrixoma from other cutaneous adnexal tumors or malignant entities. In the present case, the classical histopathological features of pilomatrixoma were clearly evident, enabling a definitive and confident diagnosis without ambiguity. Recognizing these hallmark characteristics was essential to guide the subsequent management and surgical planning. The surgical procedure was performed under local anesthesia to provide adequate pain control while allowing real-time patient cooperation and minimizing systemic risks.

A direct incision was made precisely over the lesion, providing optimal access and visibility. The dissection was conducted carefully and methodically, with meticulous attention to preserving the integrity of adjacent anatomical structures such as eyelid muscles, tarsal plate, and conjunctiva. Complete excision of the lesion was achieved with clear margins to minimize the risk of residual tumor tissue and subsequent recurrence. The importance of excising with margins cannot be overstated, as incomplete removal may lead to regrowth and necessitate further interventions.

Hemostasis was effectively controlled using bipolar cautery, which allowed for precise coagulation of small blood vessels with minimal collateral tissue damage. This technique ensured a clean surgical field and reduced the likelihood of postoperative hematoma formation. Closure of the surgical wound was performed in layers to promote optimal healing: the deep tissues were approximated using absorbable 6-0 monofilament sutures to provide stable support without the need for suture removal, while the skin was closed with fine non-absorbable 7-0 sutures to achieve minimal scarring and a satisfactory cosmetic result. The careful surgical technique, combined with the patient's good healing capacity, contributed to an excellent postoperative outcome, with preserved eyelid function and a well-camouflaged scar along the natural eyelid crease.[2,4,5]

From both functional and aesthetic standpoint, the patient's outcome was highly satisfactory. There were no postoperative complications such as infection, wound dehiscence, or hematoma, which underscores the success of the surgical approach employed. The eyelid maintained its full range of motion and structural integrity, while the resulting scar was minimal and well-concealed within the natural eyelid crease, preserving the patient's appearance without noticeable deformity. This excellent result highlights the critical importance of timely and meticulous surgical intervention, which not only addresses the lesion effectively but also safeguards the delicate balance between eyelid function and cosmesis.

Furthermore, this case serves to emphasize that, despite its rarity, pilomatrixoma should be actively considered in the differential diagnosis of eyelid nodules in adult patients. Recognizing this possibility early can profoundly influence the choice of treatment, guiding clinicians toward complete excision rather than conservative or repetitive approaches that may delay definitive care. Inclusion of pilomatrixoma in the diagnostic workup can therefore have a direct and positive impact on therapeutic planning, helping to optimize both the functional outcome and the aesthetic prognosis for the patient.

Eyelid pilomatrixoma in adults represents a rare and often elusive diagnosis, posing significant challenges within ophthalmologic practice. Its presentation is frequently atypical and can closely mimic a variety of other more common subcutaneous lesions, such as chalazion, cysts, or benign tumors, leading to frequent misdiagnosis. This diagnostic uncertainty has important implications, as it can delay appropriate management and result in unnecessary or ineffective treatments.

The case presented here vividly highlights the critical importance of maintaining a wide-ranging differential diagnosis when assessing eyelid nodules that show progressive growth over time. Given the subtle and overlapping clinical features of pilomatrixoma with other eyelid masses, relying solely on clinical examination is insufficient. Histopathological analysis remains the definitive method for confirming the diagnosis, providing clear guidance for tailored therapeutic interventions and reducing the risk of recurrence.

Early recognition followed by complete surgical excision with clear margins has been shown to be the most effective treatment strategy, as demonstrated by the excellent postoperative results and absence of tumor recurrence in this case. This experience reinforces the necessity for ophthalmologists and clinicians involved in eyelid lesion management to be well-informed about pilomatrixoma, despite its rarity, and to include it in the differential diagnosis of eyelid masses in adult patients. Such awareness ensures timely diagnosis, appropriate treatment, and optimal functional and cosmetic outcomes.

AUTHOR'S CONTRIBUTION

"Budib CL contributed to the conception and design of the case report, analysis and interpretation of the results, drafting and critical revision of the manuscript content. Ferreira GO and Mucci MMF contributed to data analysis and interpretation, and drafting and critical revision of the manuscript content. Barboza GNC, Barboza MNC, and Minalez AGT contributed to the critical revision of the manuscript content. All authors approved the final version of the manuscript and are responsible for all aspects of the work, including ensuring its accuracy and integrity."

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