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(CASE REPORT)

Rare Case of Pilomatricoma of the Leg in a Young Patient: A Case Report

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Abstract

Pilomatricoma, a benign neoplasm originating from the hair follicle, typically occurs in the head and neck region. However, its occurrence in the leg is rare. We present a case of a young patient with a palpable mass on the leg. Physical examination revealed a 2 cm firm, non-tender subcutaneous nodule. Imaging studies (ultrasound and MRI) confirmed the diagnosis of pilomatricoma. The patient underwent successful surgical excision under general anesthesia, achieving clear margins. Histopathological examination demonstrated characteristic features, including basaloid cells, ghost cells, and calcifications, with no evidence of malignancy. Regular follow-up visits showed no recurrence several months postoperatively.

Pilomatricoma is typically found in the head and neck region, making its occurrence in the leg extremely rare. Differential diagnosis includes lipomas, dermoid cysts, and neurofibromas. Radiological imaging, particularly ultrasound, aids in preoperative diagnosis by demonstrating characteristic features. Surgical excision with clear margins is the mainstay of treatment, ensuring complete removal and reducing the risk of recurrence.

We report a rare case of pilomatricoma of the leg, highlighting the importance of considering this diagnosis even in uncommon anatomical locations. Surgical excision with clear margins resulted in successful management without recurrence. Awareness of atypical presentations of pilomatricoma is crucial for accurate diagnosis and appropriate management in clinical practice.

Keywords: Pilomatricoma; Leg; Hair follicle tumor; Benign skin neoplasm; Surgical treatment; Case report

1. Introduction

Pilomatricoma, also known as Malherbe's mummified epithelioma, is a relatively uncommon and benign neoplasm that arises from the hair follicles of the skin. It accounts for less than 2% of all primary cutaneous tumors [1]. While pilomatricoma predominantly affects the head and neck region, its occurrence in the extremities is rare. This tumor often presents as a subcutaneous nodule and can be easily overlooked or misdiagnosed as other cutaneous lesions.

The objective of our research is to comprehensively investigate the diagnostic and therapeutic aspects associated with pilomatricoma by presenting a detailed clinical case study. By documenting and analyzing the clinical presentation, radiological findings, histopathological features, and treatment outcomes of a specific case, we aim to enhance our understanding of this pathology and contribute to its management and treatment. The in-depth exploration of this rare case will provide valuable insights into the atypical presentation of pilomatricoma in an uncommon anatomical location and aid in accurate diagnosis and appropriate management strategies.

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By emphasizing the distinctive features and diagnostic and therapeutic challenges of pilomatricoma occurring in an unusual anatomical location, this study seeks to enrich the existing literature and support improved clinical decision-making in comparable cases.

2. Clinical case

We present the case of a 35-year-old female patient who presented with a ten-month history of swelling on the outer edge of her right leg. The patient sought medical attention due to the concern about the persistent swelling. Upon clinical examination, a nodular swelling measuring two centimeters in diameter was observed. The swelling was firm, painless, and adherent to the skin but mobile in relation to the deep plane. The surrounding skin appeared normal (Figure 1).

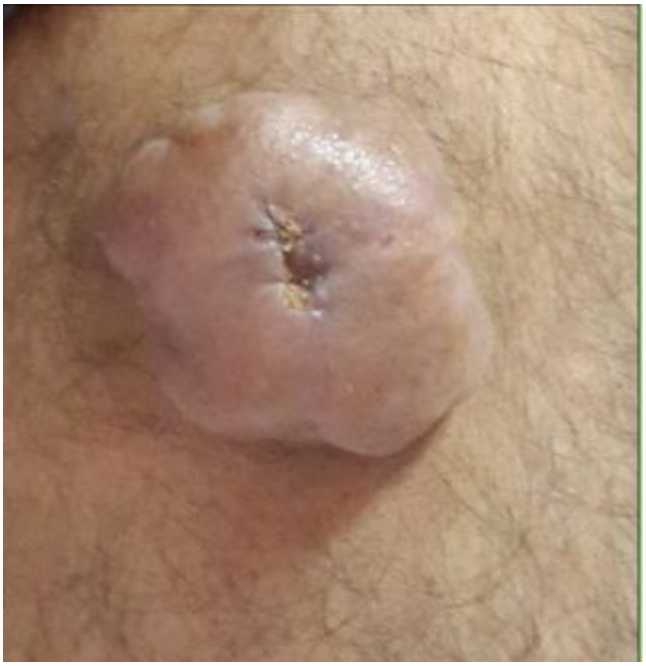

Figure 1 Clinical aspect of the pilomatricoma

Lymph node examination revealed no locoregional adenopathy, and the rest of the clinical examination was unremarkable. X-rays of the leg revealed soft tissue calcification, and further imaging scans showed a well-defined calcific hyperdensity. The patient subsequently underwent surgical biopsy, which confirmed the diagnosis of pilomatricoma.

Total excision of the tumor was performed under local regional anesthesia. Macroscopically, the tumor appeared as an encapsulated, indurated nodule measuring five centimeters in its longest axis. Histopathological examination revealed foci of keratinization and calcification, along with sheets of basaloid and mummified cells. The cytoplasmic boundaries of the cells were clearly visible, and the nuclei exhibited a distinct imprint (FIGURE 2)

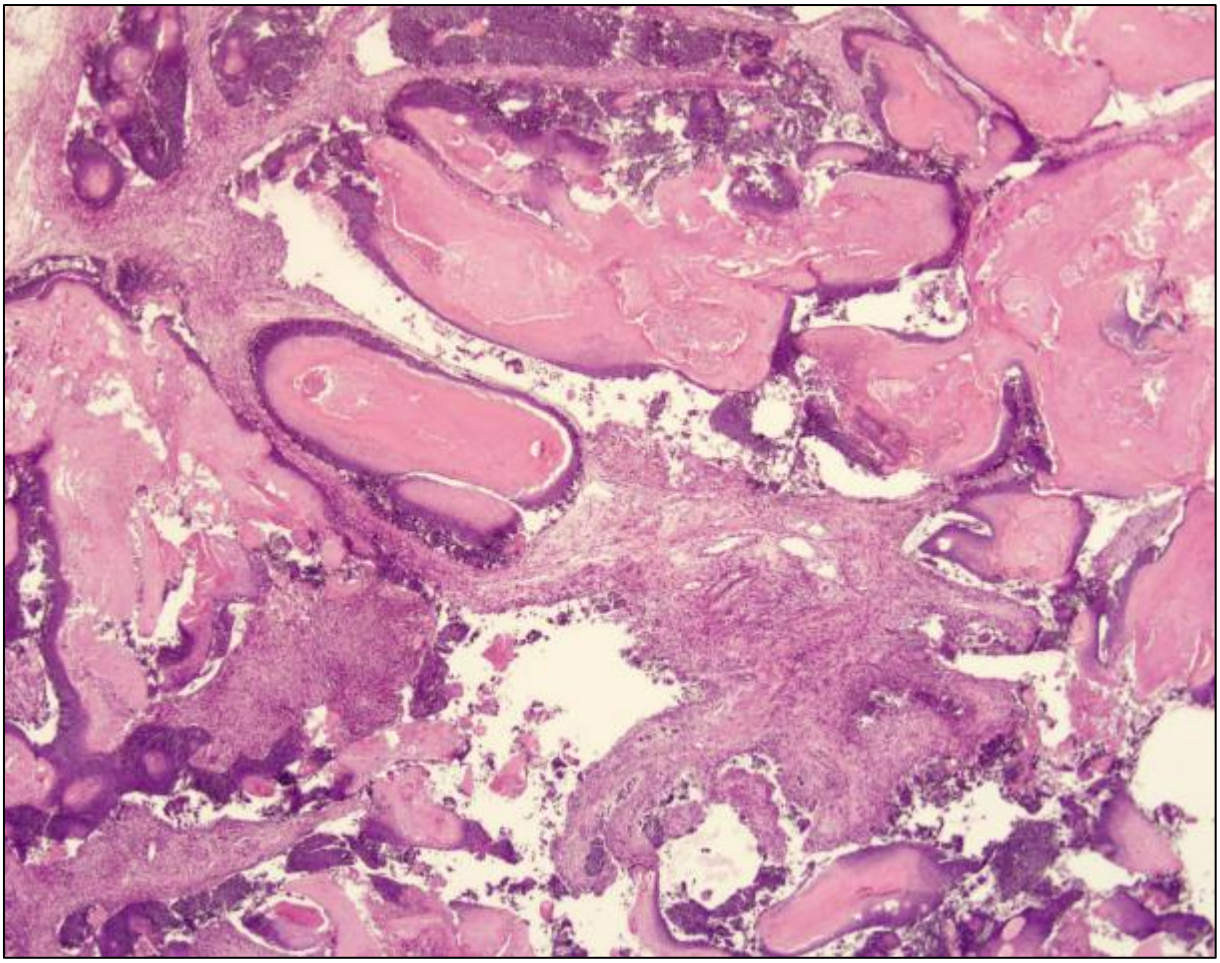

Figure 2 Histopathological features showing foci of keratinization and calcification with sheets of basaloid and mummified cells

The diagnosis of pilomatricoma was confirmed. At the 18-month follow-up, no recurrence of the tumor was observed.

3. Discussion

Pilomatricoma, also known as Malherbe's mummified epithelioma, is a benign calcified tumor originating from the hair follicle matrix [1, 2]. It typically presents as a small, solitary, subcutaneous nodule, commonly observed in the head and neck region during the first two decades of life [1, 2]. In our case, the patient presented at an atypical age of 35, and the tumor was located in the lower limb, which is a rare occurrence [4-7].

The diagnosis of pilomatricoma is primarily based on histopathological examination, which reveals characteristic features such as foci of keratinization, calcification, and sheets of basaloid and mummified cells [2]. Immunohistochemical studies have confirmed the origin of pilomatricoma from the cells of the pilar matrix [2]. In our case, the histopathological findings were consistent with pilomatricoma, confirming the diagnosis.

Pilomatricoma can exhibit various clinical forms, including familial forms associated with systemic diseases such as myotonic dystrophy and Gardner's syndrome [9]. Multiple localizations are common in these cases [7, 10]. However, our case was not associated with any known systemic diseases or multiple localizations.

The clinical presentation of pilomatricoma can vary, with some cases showing ulceration or anetodermal changes in the surrounding skin [8, 10]. In our case, the surrounding skin appeared normal. The "tent sign," described as a flat surface separate from the rest of the tumor, is a characteristic feature of pilomatricoma, but it was not observed in our patient [8].

Imaging studies, such as ultrasound, may aid in the evaluation of pilomatricoma, showing a well-defined subcutaneous mass with a "target-like" appearance and evidence of calcification [4, 10]. In our case, X-rays revealed calcification of the tumor, providing additional diagnostic information.

Differential diagnosis of pilomatricoma includes other cutaneous lesions, such as squamous cell and pilar cysts. Histological examination and immunohistochemical studies can be valuable in distinguishing between these entities [9.10]. Malignant pilomatricoma or trichomatric carcinoma, although rare, should also be considered in the differential diagnosis due to their aggressive nature [10].

The possibility of carcinomatous transformation in pilomatricoma remains controversial, with reported cases of coexistence of benign and carcinomatous forms within the same patient [5]. However, the overall prognosis of pilomatricoma is favorable, and complete surgical excision is curative with a low risk of recurrence [5-8]. In our case, the patient remained recurrence-free at the 18-month follow-up.

In conclusion, this case highlights an atypical presentation of pilomatricoma in an uncommon anatomical location. Accurate diagnosis through histological examination is crucial to differentiate pilomatricoma from other cutaneous lesions and to exclude malignant forms. Surgical excision remains the primary treatment modality, resulting in a favorable prognosis with low recurrence rates. Further research is needed to explore the underlying pathogenesis and potential prognostic factors associated with pilomatricoma.

4. Conclusion

Pilomatricoma is a benign cutaneous tumor that typically presents as a solitary lesion, most commonly found in the cervicofacial region and more frequently observed in females. Multiple lesions and involvement of the limbs are rare. Accurate histological diagnosis is essential to differentiate pilomatricoma from its malignant counterpart. The primary treatment approach is surgical excision, which is crucial for preventing recurrence.

Compliance with ethical standards

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Disclosure of conflict of interest

All authors have participated and agreed to the study of this case and have agreed to it publication.

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No source of funding took part by any mean to our case report.

Statement of informed consent

Written informed consent was obtained from 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.

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