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Pilomatrix Carcinoma of the Lumbar Skin: A Case Report With Updated Literature Review

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In accordance with local regulations, separate institutional ethics committee approval was not required for this single anonymized case report.

Informed Consent
Written informed consent was obtained from the patient for publication of this case report and the accompanying clinical and intraoperative images.

Conflict of Interest
The authors declared no conflicts of interest.

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Data Availability
Data supporting this case report are available from the corresponding author upon reasonable request, subject to patient confidentiality and institutional requirements.

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Abstract

Pilomatrix carcinoma is an extremely rare malignant adnexal tumor originating from hair matrix cells. It represents the malignant counterpart of pilomatrixoma and is characterized by locally aggressive behavior, a high rate of local recurrence, and potential for regional and distant metastasis. Most reported cases involve the head and neck region, whereas truncal involvement is uncommon. We present a case of pilomatrix carcinoma arising in the lumbar skin of a 52-year-old male patient who presented with a slowly enlarging subcutaneous mass. Histopathological examination revealed infiltrative growth of atypical basaloid cells, shadow cells, marked cytological atypia, and increased mitotic activity. The patient underwent wide local excision with sentinel lymph node biopsy, which showed no metastatic involvement. Final pathology of the excision specimen confirmed negative surgical margins, and the patient remained recurrence-free, with no nodal or distant metastasis, after 18 months of follow-up. This case highlights the importance of accurate histopathological diagnosis and supports wide local excision with adequate margins as the primary treatment approach for pilomatrix carcinoma.

Keywords: pilomatrix carcinoma, hair follicle tumors, wide local excision, sentinel lymph node biopsy, skin neoplasms

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Introduction

Pilomatrix carcinoma is a rare malignant tumor arising from the hair follicle matrix and represents the malignant counterpart of pilomatrixoma [1]. Since its initial description, it has been reported mainly as single case reports or small series; a recent review identified only 52 cases published between 2012 and 2022 [1, 2]. Clinically, pilomatrix carcinoma often presents as a firm, slow-growing dermal or subcutaneous mass and is frequently misdiagnosed before histopathological evaluation [2, 3]. Owing to its rarity, standardized treatment guidelines have not been established; however, wide local excision with negative margins is generally recommended, and increasing attention has been given to sentinel lymph node biopsy and adjuvant radiotherapy in selected cases [3-5].

We report this case because of its uncommon lumbar (truncal) location and to illustrate a practical management decision: after an initial excision revealed an involved deep margin and aggressive histological features, wide local excision was combined with lymphoscintigraphy-guided sentinel lymph node biopsy because of the unpredictable lymphatic drainage of truncal skin tumors. For this updated narrative (non-systematic) review, the literature was searched in PubMed/MEDLINE, Scopus, and Web of Science using the terms “pilomatrix carcinoma”, “pilomatrical carcinoma” and “malignant pilomatrixoma” covering the period from January 2000 to January 2025. Reports and reviews relevant to clinicopathological features, surgical margins, sentinel lymph node biopsy, and adjuvant therapy were prioritized.

Case Presentation

A 52-year-old male patient presented with a firm subcutaneous mass in the left lumbar region that had been slowly and steadily enlarging for approximately 2 years, without recent acceleration in growth. He had no history of pain, bleeding, ulceration, or discharge from the lesion. His medical history was notable only for hypertension. He had no immunosuppression, no personal or family history of malignancy or skin cancer, and no chronic sun or occupational exposure. On physical examination, the lesion was non-tender, measured approximately 3 cm in diameter, and showed minimal fixation to the underlying tissue, with no overlying skin ulceration or discoloration. There was no palpable regional lymphadenopathy. The lesion was clinically considered a benign epidermal cyst and was therefore excised as a presumed benign lesion.

An initial excisional biopsy revealed histopathological features consistent with pilomatrix carcinoma. Microscopic examination demonstrated irregular islands of atypical basaloid cells admixed with eosinophilic shadow cells, accompanied by a prominent foreign body giant cell reaction (Figure 1A). Additional findings included focal tumor necrosis (Figure 1B), marked cytological atypia with nuclear enlargement and eosinophilic macronuclei (Figure 1C), and brisk mitotic activity with atypical mitotic figures (Figure 1D). The tumor showed infiltrative growth with focal extension into the subcutaneous adipose tissue. No lymphovascular or perineural invasion was identified. The lateral (peripheral) surgical margins were free of tumor, whereas the deep surgical margin was involved; the maximum tumor diameter was 3 cm.

Immunohistochemistry was not performed because the diagnosis was established on the basis of characteristic morphological features, including basaloid cells admixed with shadow cells and abrupt keratinization. Given the rarity of the tumor and its morphological overlap with other adnexal neoplasms, the diagnosis was confirmed by a second pathologist.

Staging with positron emission tomography demonstrated no evidence of distant metastatic disease. Ultrasonography of the axillary and inguinal regions revealed no suspicious lymph nodes. Given the aggressive histopathological features and the involved deep margin, further surgical management was planned.

Preoperative lymphoscintigraphy identified sentinel lymph nodes in the left axillary and left inguinal regions. Wide local excision was performed with 2 cm peripheral margins and a deep margin extending to the aponeurosis of the external oblique muscle (Figure 2A). Sentinel lymph node biopsies were performed using a gamma probe in the inguinal and axillary regions (Figures 2B and 2C, respectively). Intraoperative frozen section analysis of the sentinel lymph nodes was negative for metastatic involvement. On final histopathological examination, the wide local excision specimen contained a residual tumor focus measuring 3 mm, with all surgical margins free of tumor; permanent sections of the sentinel lymph nodes also confirmed the absence of metastatic involvement.

The surgical defect was closed primarily, and the postoperative course was uneventful. Given the negative final surgical margins and the absence of nodal metastasis and the complete excision of the small residual tumor focus, adjuvant radiotherapy was not administered. The patient was managed with close clinical and radiological surveillance. At 18 months of postoperative follow-up, the patient remained recurrence-free, with no evidence of local recurrence or nodal or distant metastasis. He was monitored by clinical examination every 3 months during the first year and every 6 months thereafter, including ultrasonographic imaging of the regional nodal basins every 6 months, under combined surgical and oncological follow-up.

Discussion

Pilomatrix carcinoma predominantly affects middle-aged to elderly individuals and demonstrates a male predominance [1, 2]. Although the head and neck region is the most common anatomical site, the trunk accounted for approximately 11% of cases in a recent literature review, making lumbar localization relatively uncommon [2].

Histopathologically, pilomatrix carcinoma is characterized by infiltrative growth of atypical basaloid cells, increased mitotic activity including atypical mitoses, cytological pleomorphism, shadow cells, and areas of tumor necrosis [3, 7]. These features are critical for differentiating pilomatrix carcinoma from benign pilomatrixoma and other entities, including basal cell carcinoma with matrical differentiation, proliferating pilomatrixoma, and squamous cell carcinoma, which may share overlapping morphological characteristics [7, 8]. Immunohistochemically, nuclear and cytoplasmic β-catenin accumulation in the basaloid cells (with negative shadow cells),

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Figure 1. Histopathological features of pilomatrix carcinoma (hematoxylin and eosin staining): (A) immature basaloid cell islands with eosinophilic shadow cells (×40); (B) focal tumor necrosis (×100); (C) marked cytological atypia with nuclear enlargement and eosinophilic macronuclei (×400); and (D) brisk mitotic activity with atypical mitotic figures (yellow arrows) (×400). Scale bars: 500 µm (A), 200 µm (B), and 50 µm (C, D).

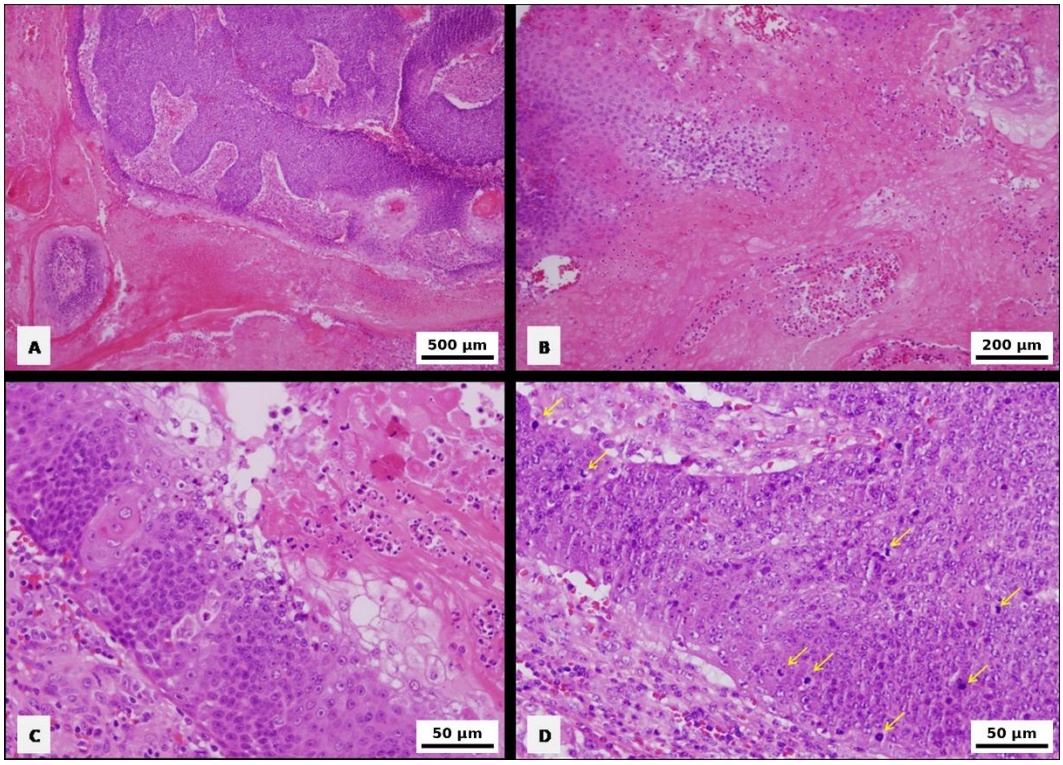
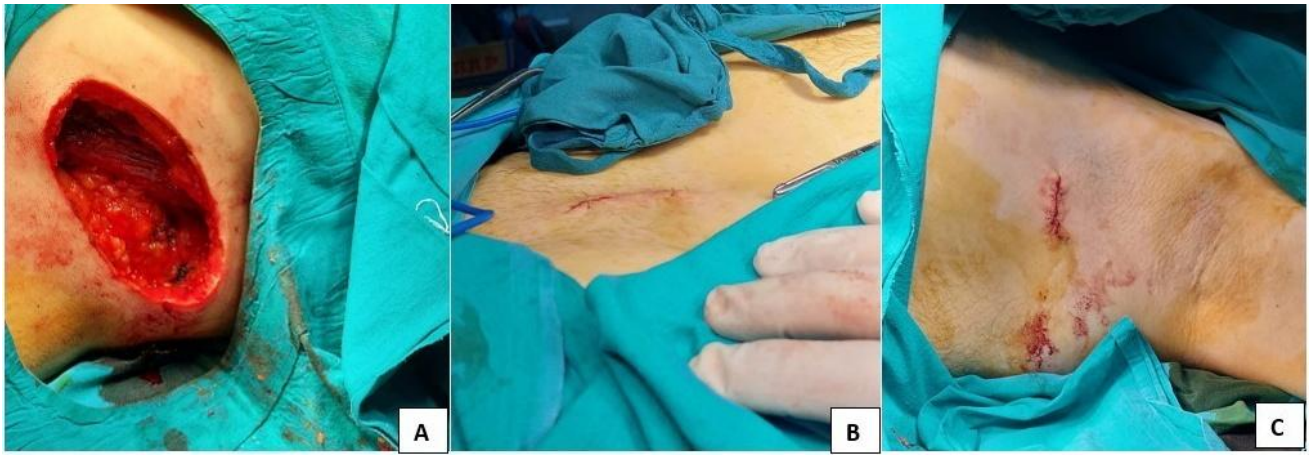


Figure 2. Surgical management of pilomatrix carcinoma: (A) wide local excision of the left lumbar skin lesion with adequate margins; (B) gamma probe-guided sentinel lymph node biopsy in the left inguinal basin; and (C) gamma probe-guided sentinel lymph node biopsy in the left axillary basin.



LEF1 and cytokeratin positivity, a high Ki-67 proliferation index, and aberrant p53 expression have been described in pilomatrix carcinoma; however, these markers support rather than confirm the diagnosis, which rests primarily on characteristic morphology [4].

Wide local excision is considered the cornerstone of treatment. Recent reviews have demonstrated that simple excision is associated with high local recurrence rates, whereas wide excision with adequate margins significantly reduces recurrence [2, 4, 6]. Surgical margins ranging from 5 to 30 mm have been proposed, reflecting the lack of consensus due to the rarity of the disease [4, 6, 9]. In the present case, 2 cm peripheral margins with a deep margin extending to the external oblique aponeurosis were chosen because of the aggressive histological features and the involved deep margin on the initial excision.

Locoregional or distant metastases have been reported in approximately 10%–17% of patients, most frequently involving the regional lymph nodes and lungs, followed by bone [1, 2, 5, 6]. In this context, sentinel lymph node biopsy is a valuable staging tool, particularly for tumors exhibiting aggressive histopathological features [5, 9]. In the present case, sentinel lymph node biopsy was undertaken because of the aggressive histology, the involved initial deep margin, and the unpredictable truncal lymphatic drainage demonstrated on lymphoscintigraphy; it provided important staging information without additional morbidity. Rather than a routine procedure, sentinel lymph node biopsy should be considered in selected high-risk patients, particularly those with aggressive histological features, margin involvement, large or recurrent tumors, or clinically uncertain drainage patterns.

The use of adjuvant radiotherapy remains controversial. Recent case series and reviews suggest that it may improve local control in cases with positive margins, recurrent disease, or nodal involvement [5, 10, 11]. Therefore, adjuvant radiotherapy may be considered in selected high-risk patients. Conversely, systemic chemotherapy has demonstrated limited efficacy, and responses are generally poor, although it may have a role in advanced or palliative contexts [10-12].

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This report has limitations. As a single case, it cannot define the optimal excision margin, establish the role of sentinel lymph node biopsy, or resolve the value of adjuvant radiotherapy in pilomatrix carcinoma. Nevertheless, it provides a clear example of individualized surgical management of a rare malignant adnexal tumor in an uncommon anatomical location, with a favorable short-term oncological outcome.

Conclusion

Pilomatrix carcinoma is a rare but potentially aggressive malignant adnexal tumor. This case underscores the need for accurate histopathological diagnosis and supports wide local excision with adequate margins as the primary treatment strategy. Sentinel lymph node biopsy may be considered for staging in selected high-risk patients, particularly those with aggressive histological features, margin involvement, or truncal tumors with unpredictable lymphatic drainage, rather than as a routine procedure. In the present case, this approach achieved negative final margins and a recurrence-free outcome at 18 months. Given the absence of standardized treatment guidelines, management should be individualized and informed by the growing body of research.

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