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An Unusual Presentation of Giant Pilomatricoma and Diagnostic Challenges: A Case Report

Fatima Zahrae Benchekroun[1,*], Mohammed Attar[2], Ilias Jerrar Oulidi[3] and Mohamed Amine Ennouhi[2]

[1]Faculty of Medicine, Pharmacy and Dentistry of Fez, University of Sidi Mohamed Ben Abdellah, Morocco, 30000
[2]Department of Plastic Surgery, Hospital Militaire Moulay Ismail, Meknes, Morocco
[3]Department of Internal Medicine, MUSC Health, United States of America

[*]**Corresponding Author:** Fatima Zahrae Benchekroun, Faculty of Medicine, Pharmacy and Dentistry Of Fez, University of Sidi Mohamed Ben Abdellah, Morocco, 30000, E-mail: fatimazahraebenchekroun@outlook.com

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Abstract

Pilomatricoma is a benign tumor frequently found in the cervicofacial region of children or young adults. It typically manifests as a subcutaneous nodule with a diameter not exceeding 3cm. In this article, we present a rare case of pilomatricoma with several unusual findings—a giant tumor measuring 11 cm in diameter—located on the trunk of a patient in his mid-40s. The combination of the patient's age, the tumor's location, and its size makes this case exceptional. Given the limited existing literature on such lesions, we believe it is crucial to shed light on atypical cases like ours.

Keywords: Giant Pilomatricoma; Unusual Presentation; Benign Tumor; Case report

List of Abbreviations: DFS: Darier-Ferrand dermatofibrosarcoma; CK20: Cytokeratin 20; HES stain: Hematoxylin-Eosin-Safranin stain

Introduction

Pilomatricoma, also known as calcifying epithelioma of Malherbe, is a rare benign tumor that demonstrates differentiation towards both the hair matrix and cells originating in the cortex [1]. This tumor is classified as an adnexal tumor because it develops at the expense of the hair follicle. It is essential not to overlook pilomatricoma, particularly when encountered in children [2]. These lesions typically exhibit slow growth and present as deep-seated, firm, small subcutaneous nodules, most commonly occurring in young patients on the upper limbs or face [1, 2].

Atypical presentations of rare tumors such as pilomatricoma can often mimic more common skin or soft tissue lesions, potentially leading to diagnostic delays if not properly identified or even misdiagnosis. Understanding these atypical presentations is vital for refining diagnostic strategies and tailoring treatment approaches in future cases to ensure appropriate and timely management. Therefore, we present a unique case of a giant pilomatricoma, measuring 11 cm in diameter, located on the trunk of an adult in his mid-40s. This case is particularly notable not only due to the patient's atypical age but also because of the unusual size and location of the tumor, which diverges from the typical presentation. This exemplifies the need for meticulous histological confirmation and consideration of the tumor's unique features to guide effective interventions.

Materials and Methods

We present a case of a mid-40s male patient with a giant pilomatricoma located on the trunk. Informed consent was obtained from the patient for publication, with all relevant information provided and understood prior to proceeding with the case report.

Results

We present the case of a patient in his mid-40s with no significant medical history, who visited our clinic with a growing mass on his trunk that had been progressing for two years. During the clinical examination, we observed a well-defined, erythematous mass with irregular and polylobed contours, along with extensions resembling crows' feet. The skin around the mass appeared stretched, resembling stretch marks, and at the center, there was an ulceration measuring 11 centimeters in length, topped by four nodules. The mass was firm, painless, superficially adherent to the skin, and mobile in relation to the deeper tissues [see Figure 1]. Considering the unusual age of onset, the location on the trunk, the large size of the tumor (11 cm) with a long axis, as well as the relatively rapid progression, the initial suspicion did not point towards Pilomatricoma. This was due to the absence of family history, previous surgeries, injuries, or traumas that could explain the condition. The patient underwent a total spindle resection and tumor biopsy, performed under local anesthesia. The resection involved a lateral margin of 0.5 cm, extending deep into the fatty tissue while respecting the aponeurosis. The wound was closed directly in two planes [see Figure 2]. Upon microscopic examination in the anatomopathological study, the tumor exhibited a proliferation of cystic structures. These structures were lined by a stratified squamous epithelium, with the inner border composed of globoid cells exhibiting abundant acidophilic cytoplasm. The lumens of the cystic structures were filled with dense acidophilic material containing calcium deposits. In other areas, the microscopic examination revealed nuclei and mummified cell sheets surrounded by a border of basaloid cells, with no signs of malignancy [see Figure 3]. Based on these findings, a diagnosis of giant pilomatricoma was confirmed. As the pathology confirmed the benign lesion, further resection was not necessary. Pilomatricomas are generally known to be non-malignant tumors, and in this case, the absence of malignant features in the microscopic examination supported the decision to forego additional resection. It's important to note that the management of pilomatricomas typically involves complete surgical excision, ensuring removal of the entire tumor and minimizing the chances of recurrence. However, given the benign nature of pilomatricomas, the absence of malignancy in the pathological examination and the tumor free margins, additional resection beyond the initial procedure was not deemed necessary in this particular case. No adjuvant treatment

was recommended. The patient had a scar assessment 15 days after the intervention and adequate healing process was noticed. The sixth month follow-up, showed a well healed up lesion, with no signs of recurrence, so no further assessment was required. The patient expressed satisfaction and no additional concerns were reported.

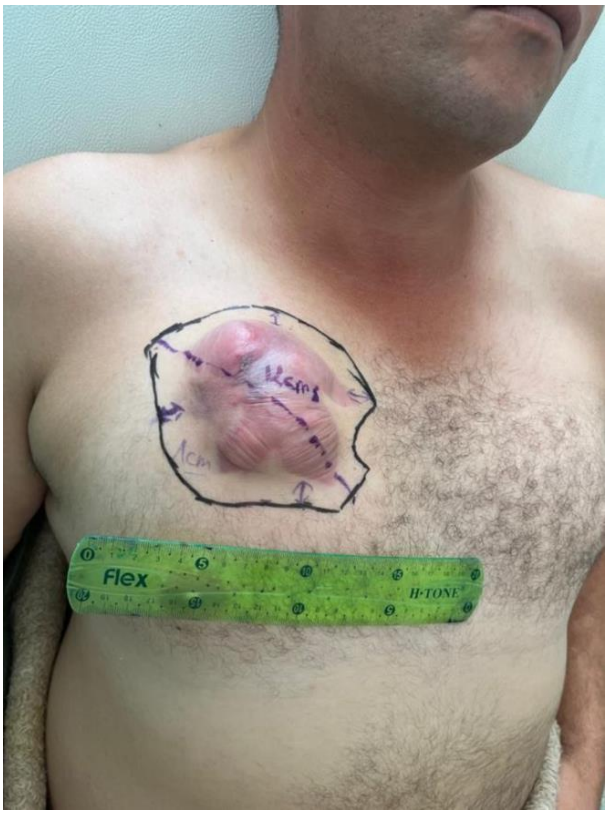

Figure 1: Image showing a giant pilomatricoma with a major axis of 11 cm, located on the trunk, with marking of a 1 cm excision margin

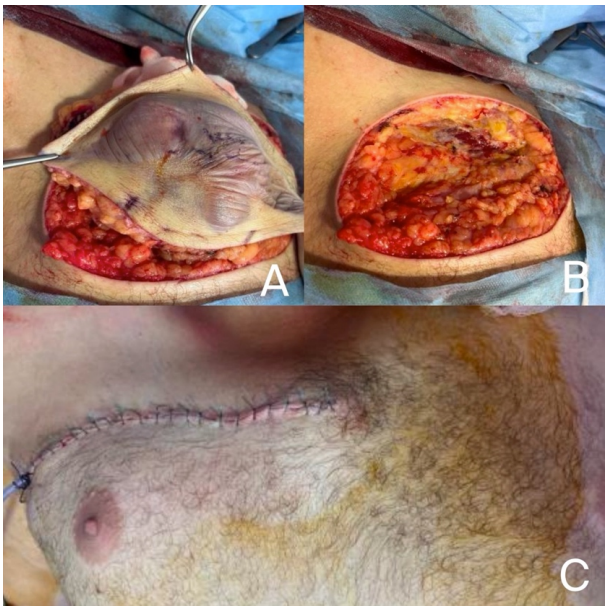

Figure 2: Images showing the surgical excision of the mass as a whole and direct suturing

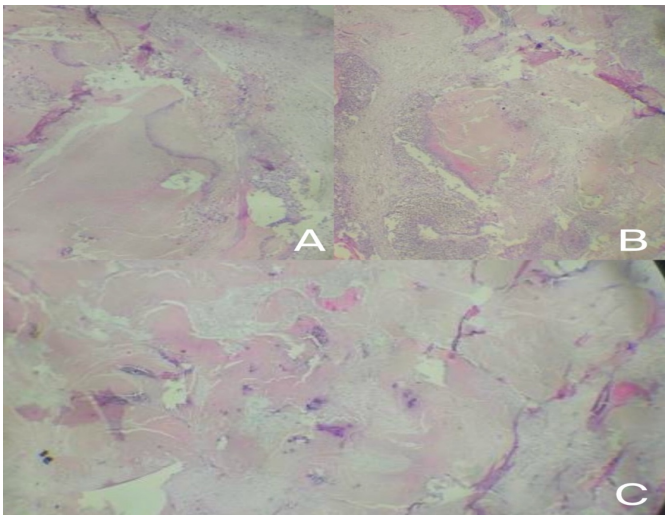

Figure 3: Anatomopathological microscopic study (HES stain): A: Tumor composed of basophilic cells phasing into eosinophilic ghost cells with adjacent fibro collagenous stroma with calcifications.

B&C: Tumor showing basophilic tumor cells phasing into eosinophilic ghost cells surrounded by fibro collagenous tissue with chronic inflammatory cells and multinucleated giant cells.

Discussion

First described by Malherbe and Chenantais in 1881, pilomatricoma, also known as calcifying epithelioma of Malherbe, is a benign tumor primarily affecting children under the age of ten [2]. Studies by Julian and Bowers, as well as Pulvermacker et al., have supported this observation, with a series of 209 cases and a large exclusively pediatric series of 89 operated pilomatricomas, respectively [3,4].

The exact cause of pilomatricomas remains not fully understood. However, researchers have observed an upregulation of the Wnt/beta-catenin signaling pathway in the normal matrix cells of hair follicles. Studies have indicated that activating mutations in beta-catenin play a crucial role in the formation of pilomatricomas and pilomatrical carcinomas. For instance, Chan et al. found that approximately 75% of the pilomatricomas they investigated exhibited beta-catenin-stabilizing activating mutations [5].

The cervicofacial region is the most common location for pilomatricomas, as shown in the study by Pulvermacker et al., where 63 out of 89 cases (71%) were located in this region, with 28% of those occurring in the jugal region. It is also more frequently observed in females, with a sex ratio of 1.5 [4]. The majority of tumors are typically less than 3 cm in size at the time of diagnosis [6]. However, our case, with a diameter of 11 cm in a male, is rare type of presentation.

Pilomatricomas generally occur during the first two decades of life and rarely beyond that period, typically between the ages of 8 and 13 [7]. They typically present as asymptomatic, round or oval, irregular, hard or firm subcutaneous nodules. The skin surrounding the lesion often appears bluish. The tumor adheres to the superficial plane but remains mobile in relation to deeper tissues. There are various clinical forms of pilomatricoma, including familial forms that are often associated with systemic diseases such as Steinert myotonic dystrophy syndrome. Involvement is frequently multiple, and familial forms are more common in such cases [8].

Pilomatricomas, a type of skin tumor, demonstrate a characteristic progression through four distinct morphological stages: The early stage manifests as small and cystic lesions. As the tumor evolves, it reaches the fully developed stage, characterized by

large and cystic lesions. During the early regressive stage, focal areas of basaloid cells, shadow cells, and lymphocytic infiltrates with multinucleated giant cells can be observed. Finally, the late regressive stage features an abundance of shadow cells, the absence of basaloid and inflammatory cells, and the possibility of calcification and ossification. [9] Based on this our case fit in the late regressive stage.

From a histological perspective, distinguishing pilomatricomas from other skin tumors involves considering several differential diagnoses. These include basal cell carcinoma, which lacks shadow cells and exhibits clefting around basaloid groups, neuroendocrine carcinoma like Merkel cell carcinoma, which lacks shadow cells but shows CK20 immunoreactivity, and proliferating trichilemmal cyst, typically characterized by large squamous cells and the absence of shadow cells [10]. Darier-Ferrand Dermatofibrosarcoma (DFS) could also be considered because of the tumor's location and clinical features. However, it is characterized by a monomorphous spindle shaped cells in a tight cartwheel pattern with little atypia and low mitotic activity, as well as neoplastic cells often infiltrating the adipose tissue surrounding it in a honeycomb pattern [11]. Additionally, pilomatrical carcinomas are differentiated by sheets, islands of proliferating atypical basaloid cells with an infiltrating border and areas of necrosis and mitoses [12].

By carefully evaluating these histological features, accurate differentiation between these entities can be achieved

Only a few exceptional cases of isolated thoracic localization in adults have been reported in the literature, making giant pilomatricomas rare. To the best of our knowledge, our case is the first to be reported thus far [13-17].

The gold standard treatment for pilomatricoma is complete surgical excision, which includes removal of the cutaneous spindle, particularly if the lesion is adherent to the dermis. This approach prevents most recurrences [18,19]. In the series by Pulvermacker et al., comprising 89 cases, no recurrences were found, while Pirouzmanesh et al. reported a recurrence rate of 1.5% in a study of 346 cases [4, 20].

It is important to note that while pilomatricoma is a benign tumor, it can rarely conceal a malignancy. In cases of malignancy, excision with a margin of 1 to 2 cm is recommended [19]. Regular surveillance is essential to detect any rare occurrences of recurrence or distant metastasis [21, 22].

Conclusion

Pilomatricoma, also known as calcifying epithelioma of Malherbe, is a rare benign tumor primarily affecting children under the age of ten.

The cervicofacial region is the most common location for pilomatricomas, with the majority of tumors being less than 3 cm in size at diagnosis.

Giant pilomatricomas, especially in adult patients, are exceptionally rare, and isolated thoracic localizations are particularly uncommon.

Complete surgical excision is the gold standard treatment for pilomatricoma, ensuring removal of the tumor and preventing most recurrences. Regular surveillance is important to detect any rare cases of recurrence or distant metastasis.

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