Frontiers in Health InformaticsISSN-Online: 2676-7104 www.healthinformaticsjournal.com
2024; Vol 13: Issue 3 Open Access

Case Report: Ossifying Pilomatricoma Masquerading as Calcified Sebaceous Cyst Causing Diagnostic Challenge- A Rare Case Report Histological Insights into Ossifying Pilomatricoma

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Cite this paper as:
Ajitha R, Mary Lilly S and Jeevprita M S J (2024). Case Report: Ossifying Pilomatricoma Masquerading as Calcified Sebaceous Cyst Causing Diagnostic Challenge- A Rare Case Report Histological Insights into Ossifying Pilomatricoma. *Frontiers in Health Informatics,* 13(3), 7535-7541.

Authors' Contributions
JMSJ carried out concepts and design, literature search, and participated in the case study. MS did histopathological reportinh of the case and helped with concept and framing the article. AR carried out concepts and design, literature search, manuscript preparation and will stand as corresponding author as well. All the authors have read and approved the final manuscript.

Abstract
Ossifying pilomatricoma is a rare, intriguing variant of pilomatricoma, distinguished by the formation of bone-like structures in the tumor. It is uncommon and poorly documented with clinical diagnostic challenges. Pilomatricoma, or calcifying epithelioma of Malherbe, arises from hair follicle matrix cells and appears as a firm, painless, subcutaneous nodule in the head, neck and upper extremities of children and young adults. This report presents a case of ossifying pilomatricoma in a 32-year-old female, detailing its clinical presentation, histopathological features, and management. While classic pilomatricoma is characterized by calcification, the ossifying variant is distinguished by its progression to true bone formation, featuring identifiable bony structures within the tumor. Identifying ossifying pilomatricoma is crucial for accurate diagnosis and management, as it influences clinical presentation, imaging, and treatment compared to non-ossifying variants. This report aims to highlight the unique histopathological features of this rare variant to improve treatment options and patient outcomes.

Keywords: *Ossification, Benign, Skin tumor, Calcifying, Histopathology, Subcutaneous.*

INTRODUCTION
Ossifying pilomatricoma is a rare, frequently misdiagnosed uncommon variant of pilomatricoma, a benign tumor originating from hair follicle cells. It commonly manifests in the head and neck region and is diagnosed in individuals under twenty years old. Pilomatricoma can show areas of calcification and focal ossification, but genuine osseous metaplasia is infrequent. Pilomatricoma is relatively uncommon, and the ossifying subtype is even rarer, posing diagnostic challenges due to its potential to mimic other cutaneous or subcutaneous conditions. This case report aims to provide a comprehensive overview of ossifying pilomatricoma, highlighting its diagnostic challenges clinically, and the significance of histopathology in diagnosis.

CASE REPORT

A 32 year old female presented with 3 year history of painless mass over right arm. The swelling was initially smaller in size and softer, recently increased in size and harder. Physical examination showed a single hard circumscribed, mobile mass of size 3x3 cm over right forearm near the wrist (Figure 1). Skin over the lesion showed hyperpigmentation and a punctum with mild serous discharge. So a clinical diagnosis of calcified sebaceous cyst was made. The entire lesion was excised under local anesthesia. Gross examination revealed a grey brown hard tissue mass which measured 3x3x2.5 cm.(Figure 2a) External surface was unremarkable. Cut section was grey brown, hard in consistency with areas of calcification.(Figure 2b) Microscopic examination showed a nodular tumor composed of lobules of basaloid cells with abrupt keratinization along with eosinophilic cells lacking nuclei(Figure 3a& 3b) intervened by areas of calcification and bone formation.(Figure 3c & 3d) The histopathological evaluation was indicative of ossifying pilomatricoma. The patient's postoperative follow up is uneventful.

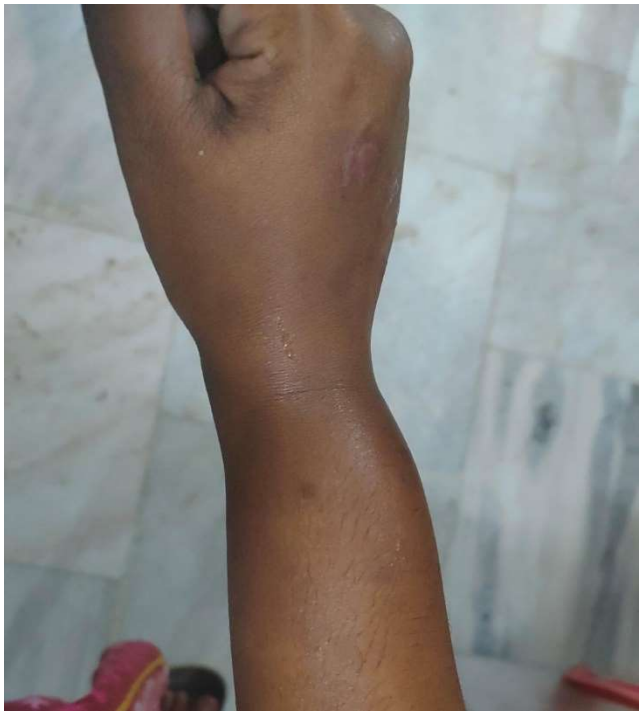

Figure 1: Ill-defined mass on forearm

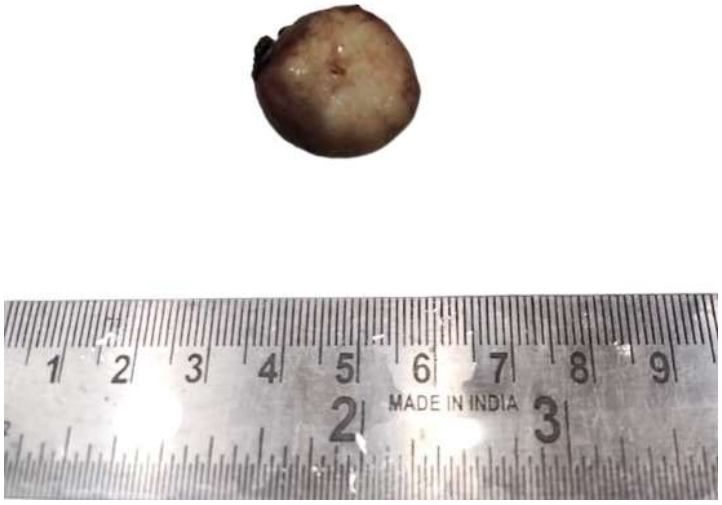

Figure 2a: gross examination showing a grey brown hard tissue mass

Figure 2b: Cut section was grey brown, hard in consistency with areas of calcification.

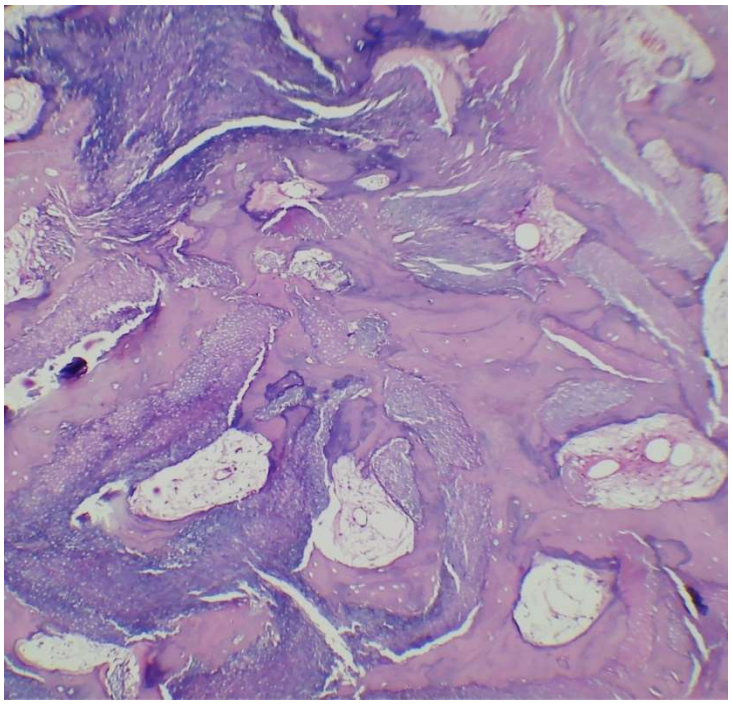

Figure 3a: Histopathology showing lobules of basaloid cells and eosinophilic cells

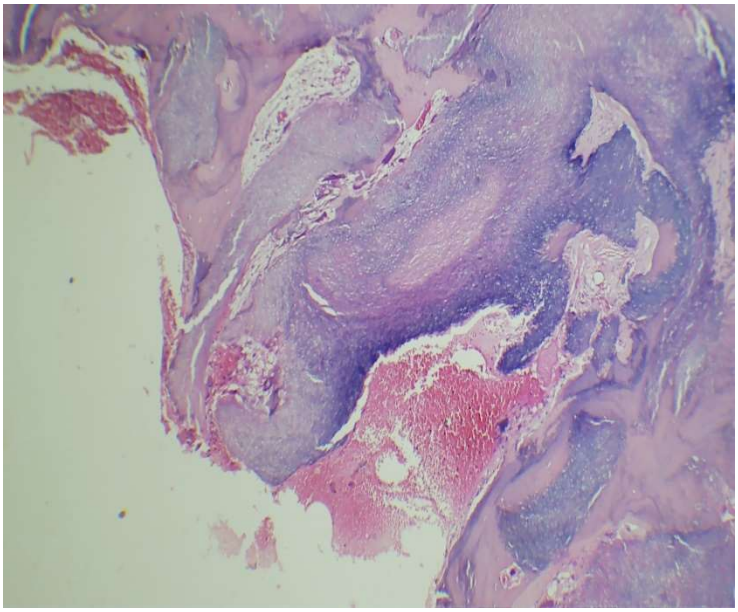

Figure 3b: lobules of cells lacking nuclei

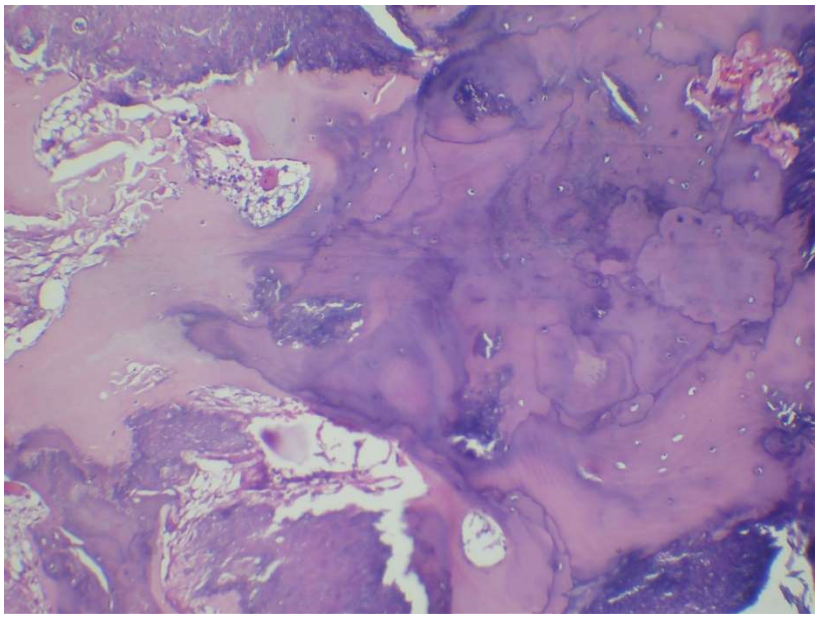

Figure 3c: areas of bone formation

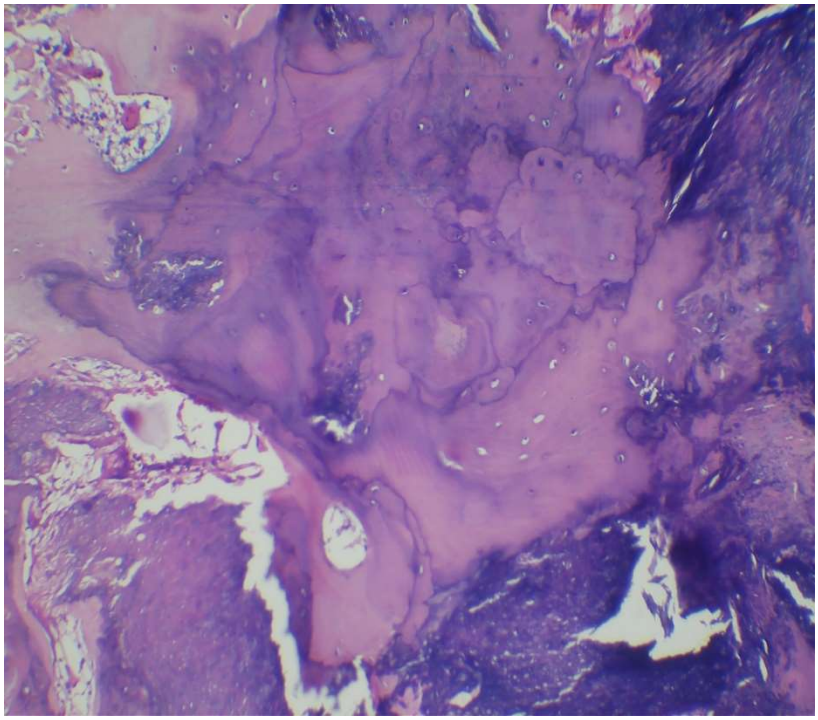

Figure 3d: areas of bone formation

DISCUSSION

Pilomatricoma, also known as calcifying epithelioma of Malherbe, is an uncommon benign skin tumor originating from matrix cells of hair follicles. Originally described by Malherbe and Chenantais in 1880 as a calcified tumor of the sebaceous glands, pilomatricoma has since been observed in numerous forms. These include bullous forms, anetodermic, lymphangiectatic, aggressive, superficial, perforating, proliferating, ossifying, cystic, pseudocystic, pigmented, acantholytic, and rare malignant variants [1]. Pilomatricoma most often appears in the first two decades of life (60%), but can occur between 4 months and 86 years. It is more common in females, with a ratio of 1.5 to 2.5 times higher than in males. In this case, the patient was a 32-

year-old female [2]. The head, neck and upper extremities are the commonly involved sites, with the tumor commonly found on the cheek, scalp and periorbital areas [3]. The tumor is covered by normal or reddened skin and ranges in size from 0.5-3 cm. When the skin over the tumor is stretched, it displays the "tent sign," characterized by multiple facets and angles, which is a distinctive feature of pilomatricoma. In our case, the size of the swelling was 3x3 cm and the skin displayed “tent sign”[4].

Although pilomatricomas have well-established features, they are often misdiagnosed. Studies reveal that the accuracy of preoperative diagnosis for pilomatricoma varies widely, from 0% to 30%. Misdiagnosis of pilomatricomas often arises from several factors, including their resemblance to cystic lesions with varying consistencies, a punctum-like appearance due to skin tethering, unusual locations, and the lack of visible calcification [4,5]. Radiologic imaging offers limited diagnostic value for pilomatricoma, and while fine needle aspiration cytology (FNAC) can help, the lack of ghost cells in the sample can lead to inaccurate results and misdiagnosis. In our case, due to clinical appearance of punctum on the lesion, it was misdiagnosed as sebaceous cyst. Pilomatricoma can rarely mimic squamous cell carcinoma (SCC) due to certain similarities in their presentation. Both can appear as firm, nodular lesions and may show areas of calcification or keratinization. In such cases, routine histopathology is crucial in making a accurate diagnosis.

The tumor has four histomorphological phases: initial cystic, mature cystic, early regressive with basaloid cells, shadow cells, and multinucleated giant cells, and advanced regression with calcification and ossification. In 1922, Dubreuilh and Cazenave described its distinctive features, including epithelial cell clusters and shadow cells. Shadow cells, which indicate abnormal keratinization, can provoke inflammatory and foreign body reactions since the body recognizes endogenous keratin as foreign.

These cells offer significant insights into the process of follicular differentiation [6]. In early lesions, basophilic cells are predominantly arranged in clusters at the periphery of the tumor. As the tumor progresses, basophilic cells lose their nuclei and become eosinophilic shadow cells. These cells frequently calcify (69-89% of cases), causing the lesion to harden, and less commonly ossify (15%) [7]. To date, there have been lesser than 15 reported instances of ossifying metaplasia with most of it occurring in the head and neck region [8]. In our study, we discuss a case of ossifying pilomatricoma of the arm which is a very unusual presentation.

The exact cause of osseous metaplasia remains unclear, but it is believed that bone morphogenic protein (BMP-2) may play a role in transforming fibroblasts into osteoblasts in advanced pilomatricomas [9]. Osseous metaplasia in pilomatricoma, as seen in this case, usually occurs in longstanding lesions and is thought to result from a prolonged fibroblastic response. Oncostatin M, secreted by activated macrophages, might facilitate the conversion of precursor cells into osteoblasts. Furthermore, the formation of calcium phosphate deposits in ghost cell clusters is thought to be stimulated by macrophage-derived osteopontin [10]. It is also proposed that macrophages arising from foreign body reactions are more influential in metaplastic ossification by driving osteoblastic differentiation and calcification more significantly than ghost cells.

Malignant transformation into pilomatrix carcinoma is a rare occurrence of this lesion commonly in the head and neck in elderly individuals. Assessing the rate of malignant transformation is challenging due to the rarity of the disease and the absence of distinct features to determine whether a malignant pilomatricoma has developed independently or evolved from a preexisting pilomatricoma. Also, spontaneous regression is never seen. So, complete Excision of the lesion is usually curative with almost no chances for recurrence, however if the lesion locally recurs it could indicate malignancy.

CONCLUSION

Ossifying pilomatricoma is a rare variant of pilomatricoma characterized by the presence of ossification within the tumor. It should be included in the differential diagnosis for patients presenting with well-defined, firm, and non-tender nodules. While it generally behaves as a benign lesion, its diagnosis can be challenging due to overlapping features with other conditions and limited radiologic and cytologic diagnostic value. Accurate diagnosis relies on histopathological evaluation, and awareness of this variant can aid in distinguishing it from other skin lesions. The condition typically involves a progression from basophilic cells

to eosinophilic shadow cells, with ossification occurring infrequently. Early and accurate diagnosis is crucial to differentiate it from other masses and avoid misdiagnosis. Although malignant transformation is rare, careful monitoring is essential to manage and address any potential complications effectively.

Consent for Publication

Written consent for the publication and any additional related information was taken from the patient involved in the study.

Declaration of Patient Consent

The authors confirm that all necessary patient consent forms have been obtained. The patient has provided written consent for their images and clinical information to be published in this journal. The patients understand that while their names and initials will not be disclosed, and every effort will be made to protect their identity.

Acknowledgements

We would like to thank the faculties of Department of Pathology for providing assistance regarding this case report.

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