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Case Report

Surgery Section

Pilomatrixoma or Sebaceous Cyst in a Middle-aged Woman: A Case Report

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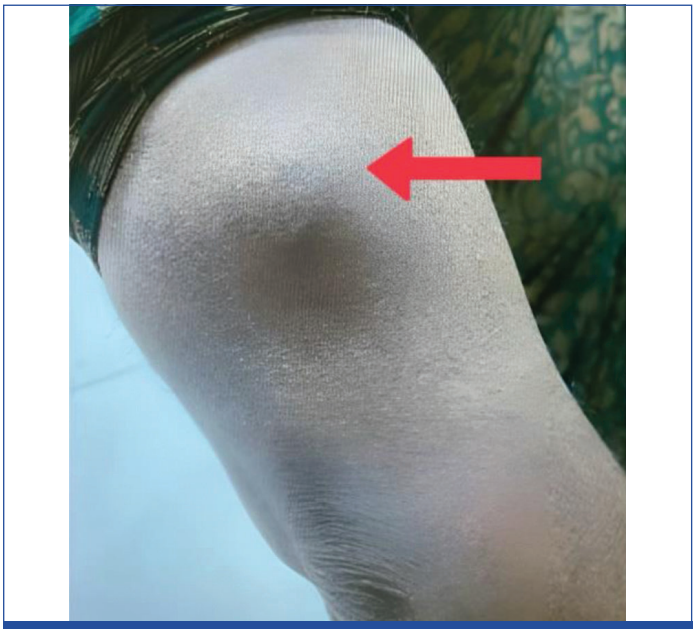
ABSTRACT

Pilomatrixoma, is a rare benign skin neoplasm originating from matrix cells of hair follicles. Frequently encountered during the first two decades of life, mostly involving the head and neck area. In this report, a woman in her late 40s presented with swelling and associated pain on her right arm. On physical inspection, a hard, non tender mass was found. A provisional diagnosis of a sebaceous cyst was made and she subsequently underwent an excision biopsy for the same. Histopathology of the specimen revealed features of pilomatrixoma. Pilomatrixoma can be confused with other benign skin masses encountered frequently in clinical practice like sebaceous cysts, especially in uncommon age groups and locations. Preoperative diagnosis is difficult to establish and a definitive diagnosis can be made only after histopathological examination. Diagnosis is through histopathology and a thorough understanding of the histological features of other cutaneous conditions is essential.

Keywords: Benign skin tumour, Epithelioma, Hair follicles, Histopathology, Neoplasm

CASE REPORT

A 49-year-old female presented to the surgical outpatient department with a conspicuous swelling located on the lateral aspect of her right arm [Table/Fig-1], which had gradually increased in size over the past year. Her chief complaints were primarily pain, though there was also a mild cosmetic concern. The patient reported experiencing pain upon touching or exerting pressure on the swelling. She denied any history of trauma, insect bites, limited range of motion, or unexplained weight loss. She did not had any significant past, personal, or family medical history of relevance.



[Table/Fig-1]: Image demonstrating swelling over lateral aspect of right arm (indicated by red arrow).

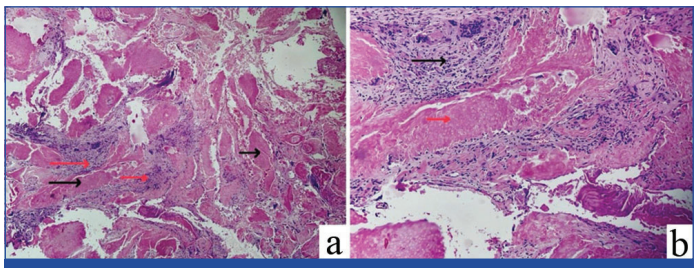
The patient's vital signs, including pulse rate, blood pressure, body temperature and oxygen saturation, exhibited no aberrations from the expected norm. During the physical examination, a solitary swelling measuring approximately 3×3 cm was identified on the lateral aspect of her right arm, just above the elbow. The skin covering the swelling and its surrounding area appeared unremarkable, with no discernible punctum. On palpation, there was minimal tenderness on the swelling, which displayed a firm consistency. Notably, the skin overlying the swelling could not be pinched and there was an absence of local temperature elevation. There was no evidence of fixation to underlying muscle or bone and no pulsations were discerned over the swelling.

A provisional diagnosis of a sebaceous cyst was made and an excision biopsy under local anaesthesia was planned. A comprehensive blood analysis was conducted, encompassing a complete blood count, a random plasma glucose assessment and an HbA1c measurement. Additionally, screenings for Human Immunodeficiency Virus (HIV) and viral hepatitis serology were diligently carried out. These evaluations did not reveal any significant abnormalities, with all parameters falling well within established normal ranges.

An elliptical skin incision was meticulously performed over the affected area. Intraoperatively, a well-encapsulated swelling, characterised by a palpable hardness in consistency, was discerned. The swelling, along with the overlying skin, was excised and the specimen [Table/Fig-2] was subsequently forwarded for histopathology examination. The surgical site was closed using vertical mattress sutures. The patient was given antibiotics as per protocol and discharged with a prescription for pain relief medications.

[Table/Fig-2]: Image demonstrating swelling contents that was excised intraoperatively.

Histopathology analysis [Table/Fig-3a,b] definitively confirmed the presence of a dermal tumour characterised by islands of basaloid cells, ghost cells and focal dystrophic calcification. Additionally, intervening stromal tissue exhibited the presence of histiocytic giant cells. These findings unequivocally constituted the classical features of a pilomatrixoma, ultimately establishing the diagnosis.



[Table/Fig-3]: Histopathology image of Pilomatrixoma. (a) Nests and islands of ghost cells (black arrow) and intermixed inflammatory stroma (red arrow) (H&E, 40x); (b) Fibrocollagenous stroma with dense chronic inflammation (black arrow) and island of ghost or shadow cells (red arrow) (H&E, 100x).

In the postoperative period, a thorough examination of the suture site on the first day revealed a healthy appearance. After the histopathological assessment, a consultation with dermatology specialists was sought. Their recommendation was a vigilant course of observation, as the excised swelling was benign. Over the following six months, the patient underwent regular follow-up examinations, all of which consistently indicated complete healing at the suture site, with no signs of recurrence detected.

DISCUSSION

Pilomatrixoma, also known as pilomatricoma or calcifying epithelioma of Malherbe, is a rare benign cutaneous tumour arising from hair follicle matrix cells [1]. It typically occurs in the first two decades of life, predominantly in the craniofacial region and is more common in females [2]. Histologically, it presents as a well-defined lesion with islands of basaloid and ghost cells [3].

The established pathogenesis, as elucidated in the literature, implicates mutations in the beta-catenin within the WNT signalling pathway and the CTNNB1 gene [1,4]. Familial occurrences of pilomatrixoma have been documented and are often associated with specific genetic conditions such as Gardner syndrome, myotonic dystrophy and Turner syndrome [2]. Pilomatrixomas are often calcified up to 90% of the time and can cause the skin above them to stretch and feel rigid when touched. While firmly attached to the overlying skin, the tumour is non tender and can be moved within the layer of subcutaneous fat [5]. A preoperative diagnosis can be made by combining clinical characteristics with different imaging techniques, such as Computed Tomography (CT), Magnetic Resonance Imaging (MRI) and ultrasonography.

Two diagnostic indicators that aid physicians in identifying a pilomatrixoma are the "tent sign," in which the skin hangs over the tumour's calcifications, produce a tent-like structure and the "teeter-totter sign," in which the contralateral edge of the tumour protrudes when one edge is pressed [5]. In this case, the tent sign was observed; however, the teeter-totter sign was not prominently noted during the examination. The most common diagnostic method, ultrasound, shows a distinct, ovoid, hypoechoic mass at the dermis-subcutis junction with a hypoechoic ring [1]. On T1-weighted images, MRI shows homogenous or inhomogeneous hypointense lesions, frequently with ring-like enhancement on postcontrast images, whereas contrast-enhanced CT shows a well-defined, distinct subcutaneous mass with variable degrees of calcification [1,4]. Given the well-encapsulated nature of the swelling, suggesting a possible benign lesion, a Fine Needle Aspiration Cytology (FNAC) was not performed preoperatively. Additionally, no imaging studies (USG/CT/MRI) were conducted, as clinical findings were indicative of a superficial lesion without joint involvement.

Histopathologically, four morphologic phases in the development of pilomatrixoma are described by Kaddu S et al., [6]. In the early stage of lesions, cystic formations are characterised by basaloid and squamous epithelium lining them, with minimal to no calcifications present. Fully grown lesions feature edges lined with basaloid epithelium and contain areas of cornified material housing ghost cells. These lesions exhibit varying degrees of dispersed calcification. Early regressive lesions comprise pink hair matrix material with multinucleated giant cells and shadow cells, without apparent epithelium. Late regressive lesions consist of calcified shadow cells with minimal to no inflammatory infiltrate, along with confluently aggregated defective hair material. They lack an epithelial component [6]. Based on histopathological findings, this case falls into the "fully grown lesion" phase, as described by Kaddu S et al., characterised by basaloid epithelium at the periphery and ghost cells at the centre [6].

Pilomatrixoma's differentials encompass sebaceous cysts, squamous and pilar cysts, subcutaneous fibromas, subcutaneous calcinosis, scrofuloderma and rarely, malignant pilomatrixoma or trichomatrical carcinoma [2,7]. Sebaceous cysts, also called epidermal or epidermoid cysts, typically occur on the scalp, face, neck, trunk and limbs. They feature a central punctum and appear as smooth, dome-shaped swellings ranging from a few millimetres to several centimetres in size [8]. On ultrasound, sebaceous cyst appears as a well-defined, homogeneous hypoechoic or anechoic lesion with posterior acoustic enhancement, representing fluid or keratin content and is typically avascular, whereas Pilomatrixoma appears as heterogeneous subcutaneous lesion with hyperechoic foci and posterior shadowing due to calcifications. Histologically, these cysts are surrounded by an epithelium resembling the epidermis, accompanied by a layer of granular cells and containing laminated keratin. While usually asymptomatic, cyst rupture can trigger a foreign body reaction. They may resemble Pilomatrixoma upon partial calcification [9,10]. While malignancy transformation is exceedingly rare, it may be a concern among middle-aged to elderly patients [7].

The primary mode of treatment entails complete excision with clear margins and the prognosis is highly favourable, with a recurrence rate ranging from 0-3% [1,11]. The reappearance of the condition should, however, arouse suspicion of Pilomatrical carcinoma [3]. The clinical challenge of differentiating pilomatrixoma from common skin conditions, especially within patient populations with limited medical access, underscores the significance of its inclusion in the realm of differential diagnosis [3].

CONCLUSION(S)

Pilomatrixoma has been an exceedingly infrequent occurrence among adults. Consequently, the challenge of establishing a diagnosis often arises due to the rarity of this condition. It is difficult to preemptively identify this condition before surgery. It is vital to underscore that the definitive gold standard for diagnosis remains histopathological examination.

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