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Pilomatrixoma – Overlooked Case Series

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ABSTRACT:

OBJECTIVE: Pilomatrixoma is a slow growing benign tumour which is most commonly misdiagnosed preoperatively as sebaceous cyst. It is a subcutaneous tumour emerging from the outer root sheath of hair follicles. We report a case series of pilomatrixoma presenting in Saveetha medical college and hospital.

METHODS: The medical records of 10 patients presented with skin swelling from a single centre from April 2022 to April 2023 were retrospectively reviewed.

RESULTS: There were a total of 10 tumours from 10 patients. The mean age at excision was 28± 5.6 years. Primary sites of occurrence include head and neck region (48.14%). No familial predominance noted. In most of the cases, a differential diagnosis of sebaceous cyst was made and final diagnosis of pilomatrixoma was confirmed by histopathological analysis.

KEYWORDS: Pilomatrixoma, outer root sheath of hair follicles, skin adnexal and appendage tumours, benign subcutaneous swellings

INTRODUCTION:

Pilomatrixoma was first documented by Malherbe and Chenantais in 1880. It was also known as calcifying epithelioma of Malherbe (1). It was called so because initially it was thought to be originating from sebaceous glands. Later in 1961, its source of origin was found to be the outer root sheath of hair follicles in the dermal or subcutaneous layers (2) by Forbis and Helwig (1). Its unique histopathological features were first noted and documented by Dubreuilh and Cazenave in the year 1922. (3) Pilomatrixoma is a benign skin lesion most commonly seen in children. Some lesions are also noted in the adult age group. Thus a bimodal distribution is observed. The primary sites of occurrence were noted to be in the head and neck region. The definitive treatment includes complete surgical excision of the tumour. Rare incidence and the non-aggressive and benign nature of this tumour poses a challenge in the accurate diagnosis of this condition. Hence a thorough understanding of etiology and pathogenesis will guide to make a diagnosis of pilomatrixoma.

MATERIALS AND METHODS:

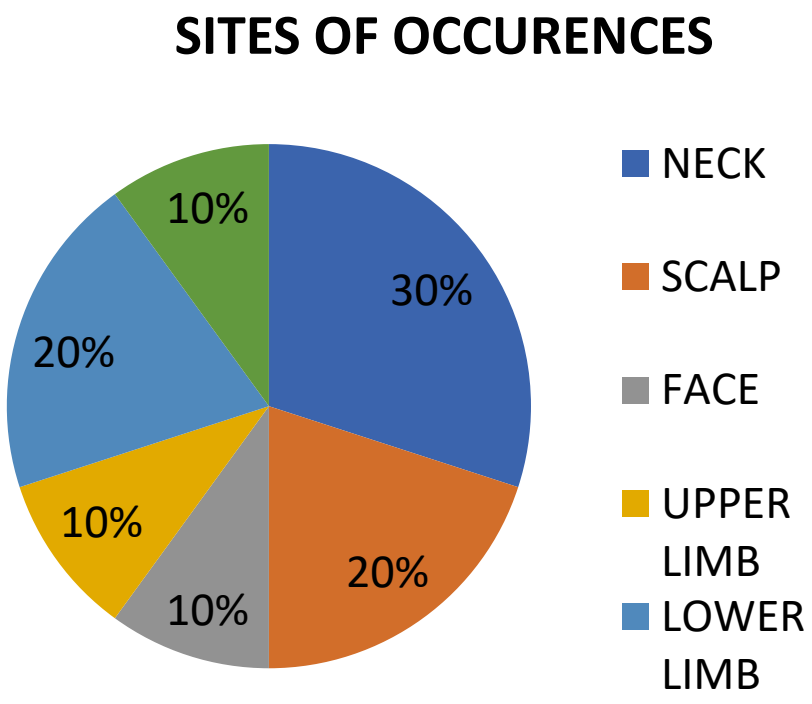
In this case series, the medical records of patients, presenting with skin lesions to the department of General surgery in our institution between March 2022 and March 2023 with histopathological examination, reported as Pilomatrixoma. The medical information contained demographic features, patient's history, clinical presentation, radiological skiagram study used, management and histopathological reports and the data was analysed retrospectively. The analysis primarily focused on key details such as gender, age, lesion size, site, and duration.

STATISTICAL ANALYSIS:

S. NO.	AGE (YEARS)	SEX	DURATION (WEEKS)	SIZE (CMS)	SITE
1.	40	M	8	4.0	SCALP
2.	29	F	4	3.4	NECK
3.	28	M	8	1.5	RIGHT CHEEK
4.	36	F	12	1.0	RIGHT THIGH
5.	60	M	6	2.5	SCALP
6.	38	F	2	2.0	NECK
7.	20	F	5	3.3	RIGHT ARM
8.	38	F	3	2.1	RIGHT UPPER BACK
9.	18	M	10	3.6	LEFT LEG
10.	21	F	3	2.2	NECK

RESULTS:

Clinical data of 10 patients were reviewed retrospectively of which 6 were females and 4 were males showing an elevated prevalence among females. The average age of occurrence was (32.8 ± 11.8) years. The mean diameter of the tumor was 2.56 ± 0.92 cms. The average duration of tumor presentation was 6.1 ± 3.31 weeks, with a time period between 2 weeks to 3 months. No positive relevant health issues was noted among the family members.The sites of occurrences of this lesion included neck (n=3), upper limb (n=2), scalp (n=2) (Fig.1), lower limb (n=2) and right cheek (n=1).



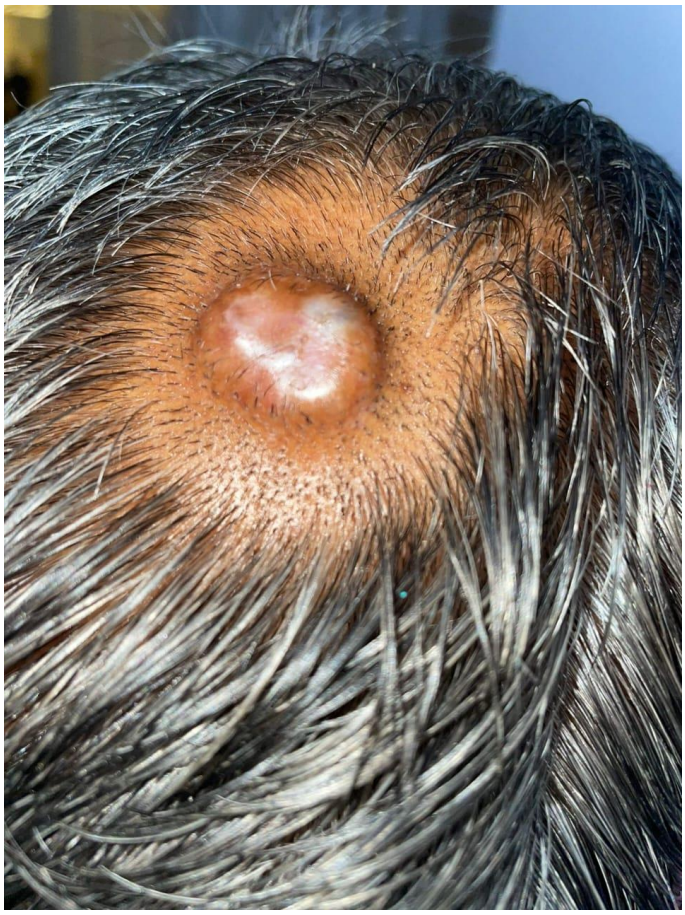


FIG 1: Clinical photo depicting a skin tumor on the scalp of a male patient measuring approximately 2x3cm with edematous skin and a punctum-like appearance.

Patients presented to the OPD with the most common complaint of skin swelling over the involved region. The swelling was more often asymptomatic, solitary, not associated with pain or local rise in temperature, with history of gradual progression in size. The swelling was cystic to firm in consistency and mobile. Slip sign negative. Skin over the swelling was pinchable. No bony indentation noted. No cough impulse seen. No complications such as acute infection, spontaneous rupture, discharge, active bleed, skin ulcerations or rapid increase in size were observed. There is no recorded history of traumatic injury.

Initially, patients underwent provisional diagnosis based on clinical examination. Radiological imaging, including X-rays (Fig.2) and ultrasound of the affected local region, was conducted. Notably, in six patients, a small punctum-like skin marking was observed, leading to misidentification as a sebaceous cyst. To distinguish the swelling from a dermoid cyst, X-rays of the involved region were performed to assess for any bony involvement or indentation. Ultrasonography revealed that all tumours were oval in shape and situated in the subcutaneous layer with no internal vascularity and few subcutaneous cystic spaces noted with in. Characteristically, the swellings appeared hypoechoic with posterior acoustic shadowing. Most lesions exhibited heterogeneity in their presentation.

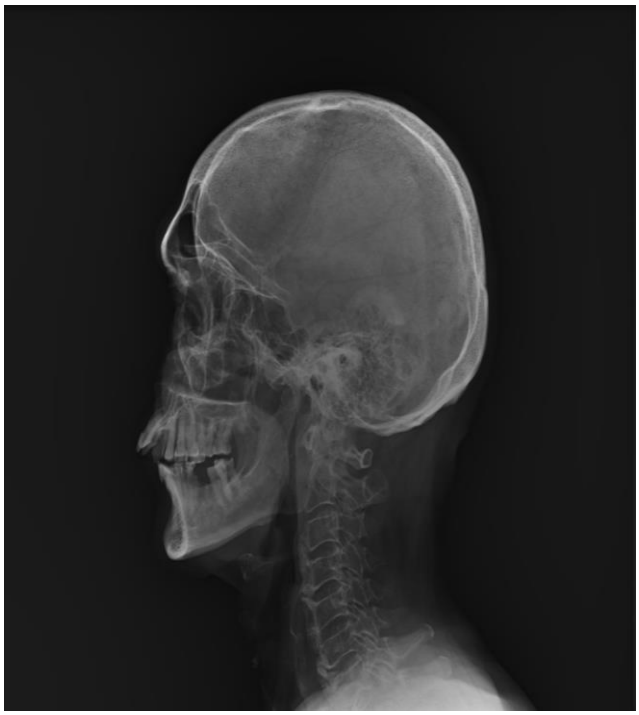


FIG 2: This image shows an X-ray of the skull lateral view taken to evaluate a patient with a scalp swelling.

In all cases, patients underwent complete excision biopsy and the diagnosis of pilomatrixoma was conclusively confirmed by a pathologist. Gross sections show grey brown globular soft tissue mass. Cut surface shows a cyst containing grey white friable material with areas of calcification. Histopathological analysis revealed epidermis with a lesion beneath composed of nests, islands and cords of basaloid cells. There are areas showing trichilemmal keratin, shadow cells, hyalinization and fibrosis. No mitosis noted. Subsequent one-year follow-up examinations post-excision indicated no recurrence.

DISCUSSION:

Pilomatrixoma is a benign neoplasm developing from the matrix cells of hair follicles. While studies suggest that trauma or insect bites may trigger its development, a CTNNB1 gene mutation, resulting in beta-catenin misregulation (4), is implicated in approximately 75% of cases.

Typically observed in the paediatric population, particularly within the first two decades of life, pilomatrixoma presents differently in our study. Here, the majority of lesions were found in younger individuals, with fewer cases noted among the elderly population. A higher prevalence was noted among females.

Pilomatrixoma typically presents as a benign skin lesion characterized by its superficial nature, slow growth, and often asymptomatic or painless behavior. The lesion is typically cystic to firm in consistency, mobile, and exhibits a lobulated surface. Skin over the swelling may appear blue erythematous (4). Pinchability of the skin over the swelling suggests its origin from the subcutaneous layer. Common sites of occurrence include scalp, neck, upper and lower extremities.

There are two distinctive pathognomic signs of pilomatrixoma. The TENT SIGN (5,6) is marked by the stretching of the skin over the tumor, displaying multiple facets and angles. Pressing

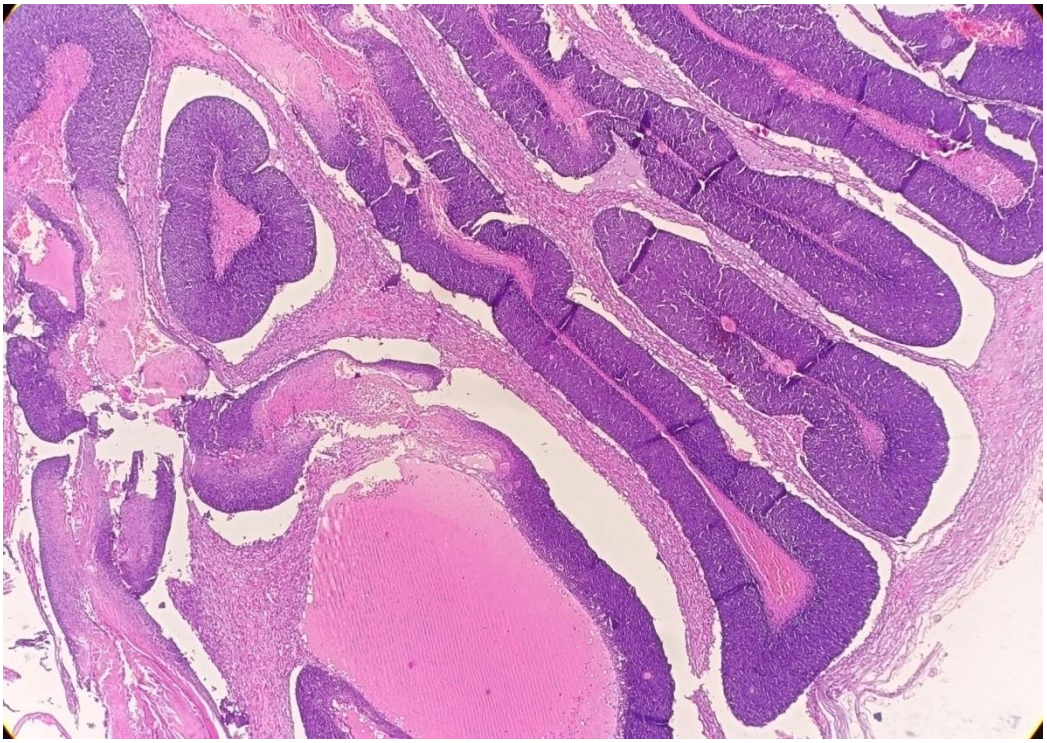
on one edge of the lesion leads to the opposite edge protruding from the skin, a characteristic known as the TEETER-TOTTER SIGN (6).

Variants of pilomatrixoma include the pseudobulbous variant, characterized by the presence of atrophic erythematous skin covering the lesion. Another variant can lead to skin ulceration and the extrusion of calcium, referred to as perforating pilomatrixomas(4). Additionally, there is the giant pilomatrixoma variant(4), defined as lesions measuring over 5 centimeters in diameter (4).

While this study did not find any lesions associated with genetic disorders, other studies have linked pilomatrixoma with various conditions such as familial adenomatous polyposis(2), Turner syndrome(2,4), neurofibromatosis type 1 (2), myotonic dystrophy(2). These pilomatrixomas typically manifest as multiple lesions.

Radiological imaging for diagnostic purposes, such as X-rays or ultrasound, may lack sufficient accuracy. Ultrasonography may aid in narrowing the differential diagnosis, with a consistent finding of hypoechogenicity in all cases, often accompanied by a hypoechoic rim representing the lesion capsule in the majority of cases and isolated or partial calcifications.

Histopathological analysis can be performed via fine needle aspiration biopsy (FNAB) or total surgical excision and biopsy. Although FNAB may help in providing a preoperative diagnosis, the cytological diagnosis of pilomatrixoma can be challenging and may lead to misdiagnosis. Therefore, biopsy post-excision remains the preferred method for ensuring an accurate diagnosis. HPE shows islands of epithelial cells (arranged in circular configuration), including shadow/ ghost cells (in the center), basophilic cells in periphery and giant cells. Basaloid cells are immature hair follicle cells with a large central core and mitosis (4). Calcification is seen in 70% of the cases. Shadow or ghost cells are cells that evolve from basaloid cells and represent dead cells which retain their cellular shape and show central unstained area that corresponds to a lost nucleus.

(A) 

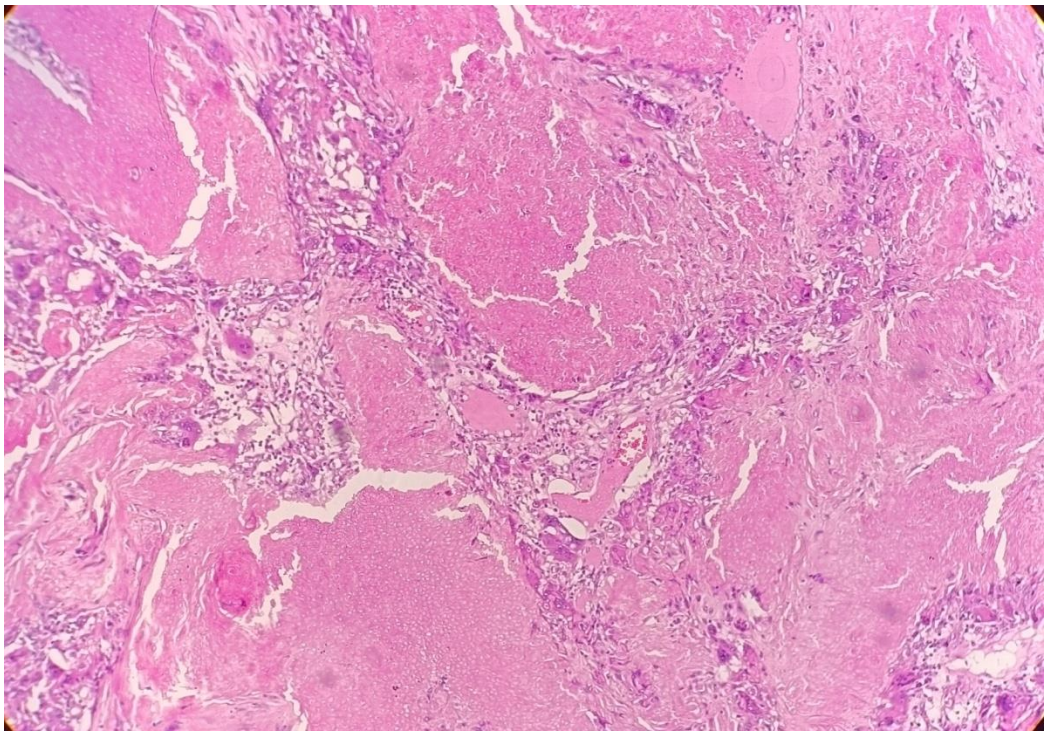
(B) 

FIG 3: H and E staining showing (A) basaloid cells and (B) ghost cells

The differential diagnosis of pilomatrixoma includes conditions such as epidermal inclusion cysts, dermoid cysts, lipoma, remnants of brachial clefts, chondroma, preauricular sinuses, osteoma cutis, foreign body reaction, ossifying hematoma, giant cell tumor, and degenerating fibroxanthoma.

The primary treatment modality involves surgical excision and biopsy of the swelling. Additional surgical techniques may include lozenge resection and avulsion. Postoperative complications can encompass surgical site infection, hematoma, wound dehiscence, hypertrophic scar or keloid formation (4). Spontaneous regression has never been observed. Malignant transformation is very rare (suspected only in cases with repeated local recurrences) (metastasis to lung, bone, brain, abdominal organs, skin, LN). Recurrence is rare (0-3%).

CONCLUSION:

Pilomatrixoma is often overlooked as a potential differential diagnosis when encountering benign skin lesions, primarily due to limited awareness of this condition. Studies indicate that the rate of preoperative diagnosis varies between from 0% and 49%. In summary, Pilomatrixoma is primarily diagnosed based on clinical assessment and further supported by additional imaging tests. The definitive diagnosis is confirmed through histopathological analysis. This study emphasizes the importance of considering pilomatrixoma as a possible differential diagnosis for benign superficial or subcutaneous lesions, particularly those occurring in the head and neck region.

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