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ORIGINAL RESEARCH

Clinical Features, Diagnostic Challenges, and Therapeutic Outcomes of 105 Pilomatricoma Cases

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Purpose: To conduct a retrospective review and analysis of clinical data on pilomatricoma over the past 9 years, with a focus on clinical characteristics, Diagnostic Challenges, and Therapeutic Outcomes.

Patients and Methods: We performed a retrospective study on patients diagnosed with pilomatricoma at our department from 2013 to 2022. Data collected from hospital and outpatient records, as well as pathological reports, included patient age, sex, onset, clinical and histopathological features, preoperative diagnosis, treatment methods, recurrence, and outcomes.

Results: A total of 105 patients were included. The primary clinical manifestation was a slowly growing subcutaneous mass. The median age at excision was 14 years, with the youngest patient being 8 months old and the oldest 71 years old. Head and neck tumors accounted for 67.6% (n=71) of cases, followed by the upper limbs (20.9%, n=22), with other sites including the back and lower limbs. The male-to-female ratio was 1:1.6. Tumor diameter ranged from 0.4 to 5.0 cm, with a mean of 1.0 cm. Only one case exhibited recurrence. Differential diagnoses included epidermoid cyst, sebaceous cyst, and fibroma, among others. The characteristic histopathological features were eosinophilic ghost cells and basophilic basal-like cells arranged in irregular strands or clusters. The preoperative diagnosis was consistent with the pathological diagnosis in only 28 cases (26.7%). The optimal treatment approach is complete surgical excision, with a low recurrence rate of 1.0%. No cases of malignant transformation were observed.

Conclusion: Pilomatricoma is a benign tumor with atypical morphology, often leading to misdiagnosis. Careful histopathological examination is crucial, and early excision demonstrates significant effectiveness in preventing recurrence.

Keywords: calcified epithelioma, pilomatricoma, benign tumor, clinical analysis, pathology

Introduction

Pilomatricoma, first described by Malherbe and Chenantais in 1880, is also known as Malherbe's calcifying epithelioma. It is a benign tumor originating from primitive epithelial germ cells that differentiate into hair matrix cells, with a low malignant transformation rate.[1,2] Literature reports indicate its incidence among benign skin tumors is 1%.[3] It typically presents as an asymptomatic, slowly growing skin mass with a firm texture, and the lesions often appear bluish in color. Pilomatricoma is frequently misdiagnosed preoperatively due to overlapping clinical features with other benign tumors, such as sebaceous cysts and fibromas. Due to its high misdiagnosis rate, pilomatricoma has gained increasing attention in recent years.[4] By reviewing the medical records and pathological reports of all outpatient and inpatient cases in our department over the past 9 years, we conducted a statistical analysis of the clinical and pathological features, as well as the treatment and outcomes of pilomatricoma. This study aims to enhance clinical understanding and improve diagnostic and therapeutic capabilities.

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Materials and Methods

A retrospective analysis was conducted on all cases of pilomatricoma that were surgically excised and pathologically diagnosed between June 2013 and June 2022. The pathological diagnoses were re-confirmed by the attending pathologists in the pathology department. Data collection included the review of hospital medical records, outpatient records, and pathology reports. The recorded variables included patient name, gender, age, clinical and histopathological presentations, preoperative diagnosis, treatment methods, recurrence, and treatment outcomes. Patients with multiple preoperative diagnoses, only the primary suspected diagnosis was included in the statistical analysis. This retrospective study was conducted at one tertiary dermatology center in China. All data were analyzed using descriptive statistics. Categorical variables were expressed as numbers and percentages, and continuous variables were described by mean, median, and range. Statistical analysis was performed using SPSS 26.0 software.

Results

Clinical Presentation

The majority of cases presented as slowly progressing, blue or red subcutaneous nodules with a firm texture and well-defined borders. Pain and tenderness were not commonly noted in the collected cases, and no ulceration or discharge was observed at the local site (Figure 1). The tumor diameter ranged from 0.4 to 5.0 cm, with an average diameter of 0.99 cm. The duration of the condition varied, with the shortest course being 7 days and the longest reaching 20 years, and the average duration was 1.1 years.

Gender and Age

Among the 105 patients, 61.0% (n = 64) were female and 39.0% (n = 41) were male, with a male-to-female ratio of 1:1.56. The median age at the time of tumor excision was 14 years, with the youngest patient being 8 months old and the oldest patient being 71 years old. The most commonly affected age group was 6 years old (n = 7).

Distribution of Locations

A total of 67.6% of the tumors were located in the head and neck region, 20.9% in the upper limbs, and 8.6% in the lower limbs. Tumors in the trunk accounted for 2.9%, all of which were located on the back, with only 3 cases in total. Further subdivision of the head and neck region revealed that 14.2% of tumors were located on the cheek, 12.4% around the ear, and both the forehead and eyebrow arches each accounted for 9.5%. Other locations each accounted for less than 9.0%, as detailed in Table 1.

Multiple Tumors

Among the 105 patients, a total of 107 tumors were excised. Only one patient had three tumors, while all other patients had a single tumor. The patient with three tumors exhibited histopathologically confirmed pilomatricoma in all lesions. Genetic testing for tumor syndromes (eg, Gardner's syndrome) was recommended but declined by the patient.

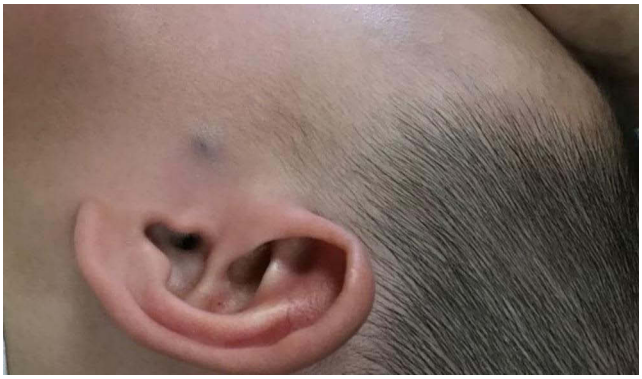

Figure 1 Hard, blue nodules located in front of the ear, with clear demarcation.

Table 1 Distribution of Skin Lesions

	Number of Cases	Percentage
Head	64	60.9%
Cheeks	15	14.2%
Around Ear	13	12.4%
Forehead	10	9.5%
Eyebrow Arch	10	9.5%
Scalp	9	8.6%
Around Eyes	4	3.8%
Around Lips	2	1.9%
Around Nose	1	0.9%
Occipital	0	0%
Mandibular	0	0%
Neck	7	6.7%
Trunk	3	2.9%
Upper Limb	22	20.9%
Lower Limb	9	8.6%

Preoperative Diagnosis

The preoperative diagnosis was primarily based on the patient's clinical presentation and the clinical experience of the attending physician, while the postoperative diagnosis was confirmed through pathological reports. The agreement between the preoperative diagnosis and the postoperative pathological diagnosis was only 26.7% (n = 28). The majority of patients, 73.3% (n=77), had alternative diagnoses, including sebaceous cysts, epidermoid cysts, fibromas, granulomas, hemangiomas, and others. The preoperative diagnoses of sebaceous cyst and epidermoid cyst accounted for 49.5% (52/105), as shown in Table 2.

Histopathological Features

The tumor is typically located within the dermis, with some cases extending into the subcutaneous tissue. Most tumors are encapsulated, appear grayish-white in color, and have a firm texture. Upon incision, the tumor is brittle and may exhibit a granular sensation. Microscopically, the characteristic features include eosinophilic ghost cells and basaloid cells arranged in irregular cords or clusters (Figure 2A). Tumor cells are generally distributed in a multilobular pattern,

Table 2 Preoperative Diagnoses in 105 Pilomatricoma Cases

Preoperative Diagnosis	Number of Cases	Percentage
Sebaceous Cyst	35	33.3%
Pilomatricoma	28	26.7%
Epidermoid Cyst	17	16.2%
Fibroma	8	7.6%
Granuloma	4	3.8%
Hemangioma	2	1.9%
Lipoma	2	1.9%
Scar	2	1.9%
Viral Wart	1	1.0%
Pigmented Nevus	1	1.0%
Nodular Hyperplasia	1	1.0%
Foreign Body Reaction	1	1.0%
Syringoma	1	1.0%
Intradermal Nevus	1	1.0%
Acne-like Carcinoma	1	1.0%

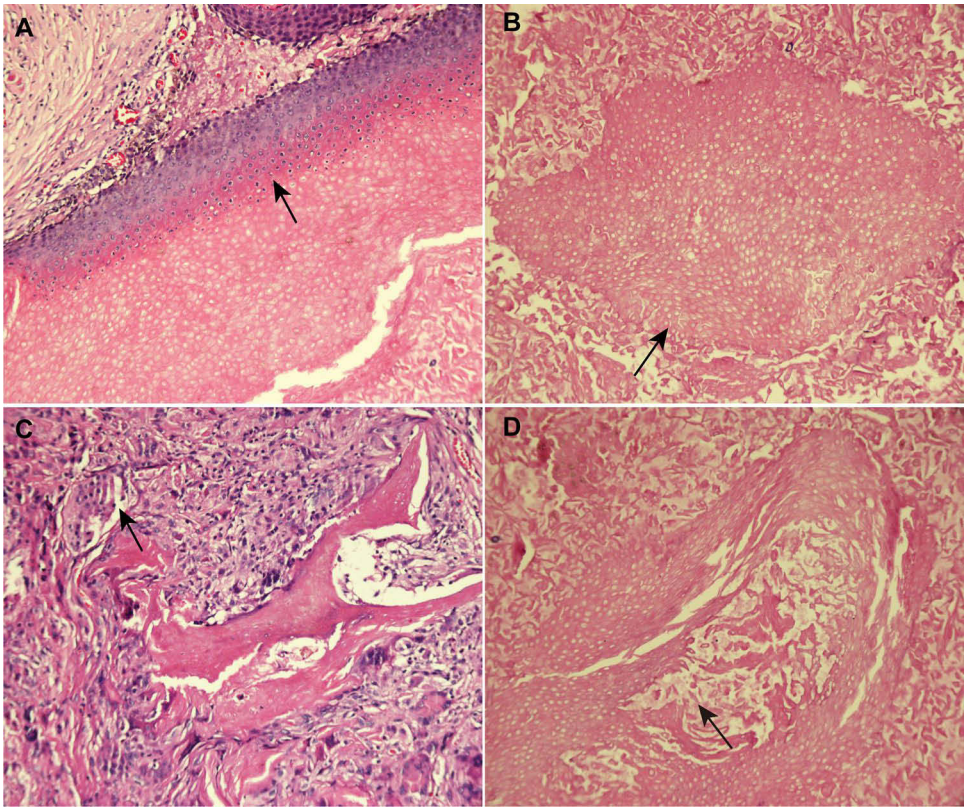

Figure 2 Histopathological features of pilomatricoma visualized by hematoxylin and eosin (H&E) staining at high magnification (×200). (**A**) A well-demarcated dermal tumor nest composed of peripheral basaloid cells with hyperchromatic nuclei and scant cytoplasm, arranged in a characteristic palisading pattern.The black arrow indicates the basaloid cell layer lining the periphery of the tumor island. (**B**) Eosinophilic anucleate ghost cells occupy the central portion of the lesion, retaining distinct cell contours but lacking nuclei, indicative of keratinization.The arrow highlights ghost cells showing progressive nuclear loss and cytoplasmic eosinophilia. (**C**) Multinucleated foreign body giant cells surround keratin debris released from ruptured ghost cells, reflecting a granulomatous reaction.The black arrow points to a foreign body giant cell, characterized by multiple nuclei within a single cytoplasmic body, adjacent to keratin deposits. (**D**) Basophilic amorphous calcified deposits are localized within ghost cell areas.The arrow marks a focus of calcification, appearing as dark-blue amorphous material among ghost cells.

occasionally surrounded by fibrous connective tissue. In the early stage of the lesion, basaloid cells predominate. These cells are small, uniform in size, with round or oval nuclei, and are typically arranged around tumor islands. As the lesion matures, ghost cells gradually become dominant, with indistinct contours, condensed nuclei, and eventual nuclear loss (Figure 2B). In some cases, infiltration of foreign body giant cells can be observed (Figure 2C). Approximately 20% of patients exhibit ossification, which usually presents as osteogenic changes or calcification (Figure 2D).

Treatment Modalities and Outcomes

All patients underwent complete surgical excision. No cases of malignant transformation were observed. Recurrence occurred in one patient (1.0%) following curettage, while wide excision cases showed no recurrence.

Discussion

The exact etiology of pilomatricoma remains unclear. Studies have suggested a genetic correlation, with conditions such as multiple tumors, myotonic dystrophy, Gardner's syndrome, xeroderma pigmentosum, and basal cell nevus syndrome accounting for over 42% of all related genetic disorders.[5] A small proportion (3.9%) of cases are attributed to trauma, such as injuries, insect bites, or surgeries.[6,7] The specific pathogenesis includes overexpression of the anti-apoptotic protein BCL-2, mutations in the CTNNB1 gene leading to activation of LEF-1 (a downstream transcription factor in the WNT signaling pathway), and the expression of hHB1 (human hair keratin basic 1). However, no studies have yet identified the precise biochemical factors contributing to the development of the disease through specific biochemical data.

According to numerous previous studies, the incidence of pilomatricoma typically follows a bimodal distribution, with the first peak occurring between the ages of 0 and 20 years, and the second peak between 50 and 65 years. The average age of onset is 4.5 years, with 90% of patients being under 10 years old. Previous reports have suggested a female predominance (male-to-female ratio of 1.1–3.3), with the head and neck region being the most common site of pilomatricoma (accounting for 40% to 77% of reported cases), followed by the upper limbs, trunk, and lower limbs.[8–11] In this study, the male-to-female ratio was 1:1.56, with 67.6% of the cases located in the head and neck area. Among the head and neck cases, the most common locations were the cheeks, around the auricle, the eyebrow arches, and the forehead. No cases were reported in the palms or soles. This distribution largely aligns with general epidemiological statistics. Recent studies across Asia have systematically summarized the epidemiological characteristics of pilomatricoma. A retrospective study conducted in China in 2020 analyzed 165 pediatric cases and reported that 63.03% of the patients were female, 93.3% were under 10 years of age, and 84.2% of lesions were located in the head and neck region, findings that are highly consistent with our study.[10] Similarly, a 2021 study from South Korea involving 59 pediatric cases of head and neck pilomatricoma found that 66.1% of cases occurred in children aged 5–14 years, with a female predominance of 59.3%.[12] In addition, a large-scale Japanese study including 1600 patients reported a male-to-female ratio of 1:1.6, with 32.1% of cases occurring in individuals aged 0–19 years, and 44.6% of lesions located in the craniofacial region.[13] Collectively, these studies confirm the high incidence of pilomatricoma in pediatric populations and its predilection for the head and neck, supporting the epidemiological trends observed in our cohort. Clinically, pilomatricomas present as slowly growing subcutaneous tumors that are firm, freely movable, and surrounded by a fibrous capsule. The overlying skin may appear blue or red, and ulceration may occasionally occur. Patients are typically asymptomatic, but a few may experience pain or itching. Most cases are solitary, with some being multiple.[14] When the skin is stretched, the tumor may appear tent-like due to flattening of the affected area. This study also did not observe any tendency for pilomatricoma to develop into malignant tumors.

The differential diagnosis of pilomatricoma is often challenging due to its variable clinical presentation and nonspecific features. It is frequently misdiagnosed as epidermoid cysts, sebaceous cysts, fibromas, lipomas, granulomas, hemangiomas, or other benign cutaneous nodules. In atypical presentations such as vesicular pilomatricoma, conditions like bullous lichen planus or drug eruptions should be considered. Lesions in the parotid region may mimic salivary gland tumors or mixed neoplasms. Histologically, while the presence of shadow cells is a key diagnostic feature, similar cells may also appear in other follicular tumors such as infundibular or trichilemmal cysts.

Consistent with these diagnostic complexities, recent studies across Asia have reported high rates of preoperative misdiagnosis. A 2023 Chinese study found that only 14.3% of 75 pilomatricoma cases were correctly diagnosed before surgery, with the majority mistaken for epidermoid or sebaceous cysts.[15] In Taiwan, a retrospective review of 179 cases reported a preoperative diagnostic accuracy of just 1.1%.[16] Similarly, a large-scale Japanese study involving 1600 cases showed a diagnostic accuracy of 48.5%, underscoring the frequent confusion with other skin tumors.[13] These findings highlight the need for greater clinical awareness and the value of adjunctive tools such as ultrasonography or dermoscopy to improve diagnostic accuracy.

The most common pathological findings include shadow cells and calcified basophilic cells. In fact, the histological appearance is variable and differs at various stages. In the early stage, there may be prominent mitotic activity, but no pathological mitotic figures are observed. This can lead to the possibility of malignancy being considered, but in reality, it is a normal phenomenon associated with the rapid growth phase of cells. In later stages, as the tumor matures, the basaloid cells transform into shadow cells, with eosinophilic cytoplasm, smaller nuclei, and enriched chromatin, and eventually, the nuclei disappear. In 80% of mature lesions, calcification is seen, with punctate basophilic calcific foci being commonly observed, and occasionally large calcified masses. Approximately 20% of patients exhibit ossification phenomena.[17,18]

Regardless of the tumor location, the best treatment method is complete surgical excision of the lesion. The recurrence rate of pilomatricoma is very low, with previous data ranging from 1.12% to 3%. When recurrences occur, it is likely due to inadequate surgical margins.[18,19] In our study, Only one case (1.0%) of recurrence was documented during a mean follow-up period of 3 years, attributed to incomplete excision, which is lower than the previous data, possibly because the surgical margin for tumor excision was extended, particularly in the extremities. For this reason, the

method of curettage within the lesion appears to be inadvisable. However, there is no unified standard regarding the size of the excision margin. Currently, there have been 125 reported cases of malignant transformation of pilomatricoma. For malignant cases, it is recommended to excise an additional 1–2 cm of tissue around the lesion,[20] and radiotherapy can also be considered in combination.

This study is retrospective and lacks a control group, making it impossible to compare the differences between curettage and excision of the lesion. Additionally, no imaging examinations or fine-needle aspiration biopsy were performed. This may be due to the variety of differential diagnoses for pilomatricoma and the attending physicians' limited knowledge of the advantages of these auxiliary diagnostic methods. Computed tomography (CT) or ultrasound can be used to differentiate calcifications and exclude vascular or sebaceous tumors. Ultrasound can also help distinguish heterogeneous echo patterns, calcification, hypoechoic borders, and posterior shadowing. These imaging techniques can assist in achieving a correct preoperative diagnosis in 76% of cases, which is significantly higher than the 33% clinical diagnosis rate.[11,20,21] In cases where the diagnosis remains unclear, fine-needle aspiration biopsy may be considered.[20,21]

In summary, through a retrospective analysis of the clinical features and pathology of 105 cases of pilomatricoma, this study aims to enhance the understanding of pilomatricoma, reduce clinical misdiagnosis, and improve treatment outcomes.

Data Sharing Statement

The data that support the findings of the study are available on request from the corresponding author.

Ethical Approval and Informed Consent

This study was approved by the Institutional Ethics Committee of the Taihe Hospital, Hubei, China. The study was conducted in accordance with the Declaration of Helsinki.

Written informed consent have been provided by the patients and their legal guardians. They have also consented to the publication of their case details.

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Disclosure

The author(s) report no conflicts of interest in this work.

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