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Pilomatricoma of the Ear Lobule in a Child Mimicking a Sebaceous Cyst: A Rare Case Report

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Abstract: ***Background: Pilomatricoma is a benign skin adnexal tumor arising from hair matrix cells. Ear lobule involvement is rare. Case Presentation: A 6-year-old girl presented with a 2 × 1 cm swelling of the ear lobule of one-month duration. The lesion was provisionally diagnosed as antibioma or sebaceous cyst. Complete surgical excision was performed. Histopathological examination confirmed pilomatricoma. Postoperative recovery was uneventful. Conclusion: Pilomatricoma should be considered in the differential diagnosis of pediatric ear lobule swellings. Histopathology remains the gold standard for diagnosis.***

Keywords: Pilomatricoma, Ear Lobule, Pediatric Tumor, Sebaceous Cyst, Antibioma

1. Introduction

Pilomatricoma, also known as calcifying epithelioma of Malherbe, is a rare benign adnexal neoplasm that arises from the hair matrix cells of the pilosebaceous unit. First described by Malherbe and Chenantais in 1880 as a calcified tumor originating from sebaceous glands, subsequent histopathological studies established its origin from primitive hair cortex cells. Pilomatricoma accounts for approximately 0.1–1% of all benign skin tumors and predominantly affects children and young adults, with a slight female predilection.

Clinically, pilomatricoma presents as a solitary, slow-growing, firm, subcutaneous or intradermal nodule, most commonly occurring in the head and neck region, followed by the upper extremities. The lesion is usually asymptomatic but may occasionally exhibit overlying skin discoloration, ulceration, or inflammatory changes. Despite its characteristic clinical features, preoperative diagnosis remains challenging, and the lesion is frequently misdiagnosed as epidermoid cyst, dermoid cyst, lymphadenopathy, or other cutaneous tumors.

To the best of our knowledge, only five pediatric cases of ear pilomatricoma have been reported in the literature so far.

Histopathologically, pilomatricoma is characterized by a biphasic population of basaloid cells and eosinophilic anucleate “ghost” or “shadow” cells, often accompanied by calcification, foreign-body giant cell reaction, and, in some cases, ossification. Molecular studies have demonstrated activating mutations in the CTNNB1 gene encoding β-catenin, implicating dysregulation of the Wnt/β-catenin signaling pathway in tumor pathogenesis.

Although pilomatricoma is generally benign, awareness of its clinical and histopathological characteristics is important to avoid diagnostic errors and ensure appropriate surgical management. Complete surgical excision remains the treatment of choice and is associated with a low recurrence rate. The present case aims to evaluate the clinical, histopathological, and therapeutic aspects of pilomatricoma and contribute to the existing literature on this uncommon skin adnexal tumor.

2. Review of Literature: Pilomatricoma

Historical Background

Pilomatricoma, formerly known as calcifying epithelioma of Malherbe, is a benign adnexal tumor originating from hair matrix cells. It was first described by Malherbe and Chenantais in 1880, who initially believed the lesion arose from sebaceous glands. Later histopathological studies demonstrated its derivation from hair follicle matrix cells, leading to the term "pilomatricoma" or "pilomatrixoma." Recent literature recognizes pilomatricoma as a relatively common benign follicular neoplasm, particularly in children and young adults.

Epidemiology

Pilomatricoma accounts for approximately 0.1–1% of benign cutaneous tumors. It occurs at any age but shows a predilection for the first two decades of life. Several studies have reported a slight female predominance. The head and neck region is the most common site, accounting for more than half of reported cases, followed by the upper extremities, trunk, and lower limbs.

Etiopathogenesis

The pathogenesis of pilomatricoma is strongly associated with mutations in the CTNNB1 gene, which encodes β-catenin, a key component of the Wnt signaling pathway. Aberrant activation of β-catenin results in uncontrolled proliferation and abnormal differentiation of hair matrix cells. Molecular biology identified CTNNB1 mutations in a significant proportion of pilomatricomas, supporting the role of Wnt/β-catenin signaling in tumor development.

There are multiple pilomatricomas which have been in association with several genetic syndromes, including myotonic dystrophy, familial adenomatous polyposis (Gardner syndrome), Turner syndrome, Rubinstein–Taybi

syndrome, Kabuki syndrome, Sotos syndrome, and constitutional mismatch repair deficiency syndrome.

3. Clinical Features

Clinically, pilomatricoma presents as a slow-growing, firm, mobile, painless dermal or subcutaneous nodule. Lesions usually measure between 0.5 and 3 cm, although giant variants have been reported. The overlying skin may appear normal, erythematous, bluish-red, or ulcerated. Characteristic clinical signs include the "tent sign," produced by stretching the skin over the lesion, and the "teeter-totter sign," where pressure on one side causes elevation of the opposite side. Despite these features, preoperative diagnosis remains difficult, with reported clinical diagnostic accuracy as low as 16%.

4. Case Report

A 6-year-old girl presented to the Department of General Surgery, Travancore Medical College, Kollam, Kerala with a swelling of the right ear lobule for one month. There is a small swelling measuring approximately 2 × 2 cm over the right ear lobule associated with erythema and warmth. Surface is smooth and edge is well defined. It is attached to skin. It is firm in consistency, fluctuant and not transilluminant. Overlying skin is normal.

She was being managed conservatively with topical applicants and antibiotics by local practitioners which yielded no results.

No previous operative history.

No familial history.

Patient was thin built and well nourished.

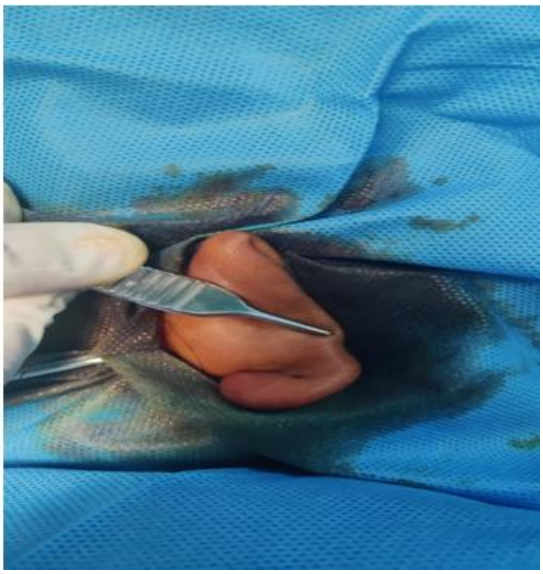
Swelling is pressed from all sides uniformly. Overlying skin cannot be pinched and its painless.

No pallor, icterus, clubbing, generalised lymphadenopathy.

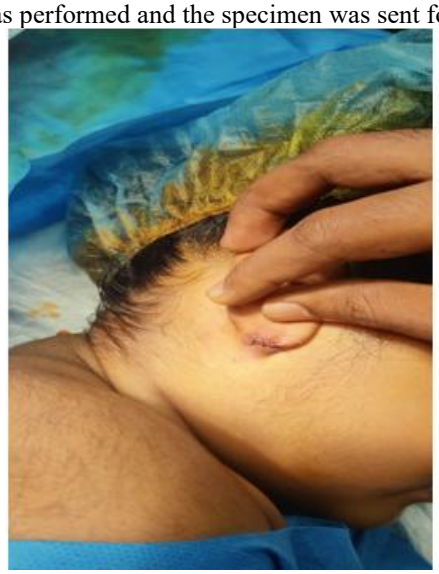
5. Evaluation

Cbc, Serology markers, aPTT, PT-INR.

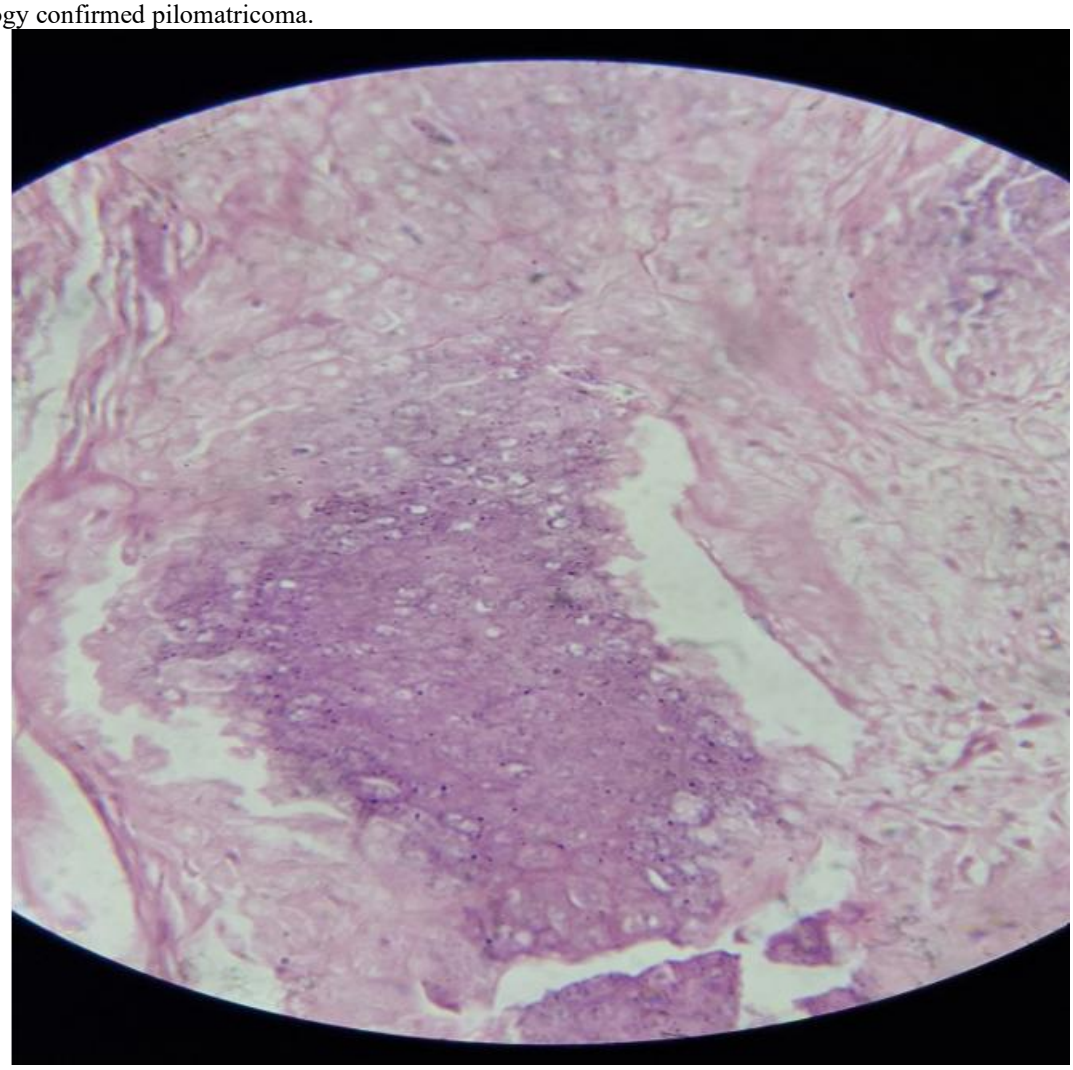
Clinical diagnosis was antibioma or sebaceous cyst.



Complete excision was performed and the specimen was sent for histopathological examination



Histopathology confirmed pilomatricoma.



6. Results and Discussion

Pilomatricoma is an uncommon benign adnexal neoplasm arising from hair matrix cells. Despite its distinctive histopathological appearance, preoperative clinical diagnosis remains challenging because of its resemblance to epidermoid cysts, dermoid cysts, lymphadenopathy, and other cutaneous lesions.

The present case involved a pediatric patient, which is in accordance with the observations of Julian and Bowers, who reported a predominance of pilomatricoma during the first two decades of life. The present case involved a female patient, which is in accordance with the slight female predominance of pilomatricoma reported in the literature.

The head and neck region was the most frequently affected site, accounting for the majority of lesions. Similar observations have been reported by Jones et al., who found that more than half of pilomatricomas occur in this anatomical location. The high concentration of hair follicles in this region may explain this distribution.

Histopathologically specimen shows fragments of neoplasm composed of islands of shadow cells with central pallor and basaloid cells exhibiting abrupt keratinization. Focal areas of calcification are also noted. Abundant fragments of keratin and keratin whorls are noted. Classical biphasic pattern composed of peripheral basaloid cells and central shadow cells. Calcification was one of the most frequent accompanying findings. These observations support the theory that pilomatricoma undergoes progressive keratinization followed by dystrophic calcification during tumor maturation.

Recent molecular studies have demonstrated activating mutations of the CTNNB1 gene, resulting in dysregulation of the Wnt/β-catenin signaling pathway. These findings provide important insights into the pathogenesis of pilomatricoma and explain its origin from hair matrix cells. Although molecular testing was not performed in the present study, the histopathological findings strongly support the diagnosis.

Complete surgical excision remains the gold standard treatment. The low recurrence rate observed in this case is comparable to that reported in the literature and highlights the effectiveness of complete removal. Malignant transformation is extremely rare and was not identified in this case.

The findings of the present case are generally in agreement with previously published literature regarding demographic characteristics, anatomical distribution, histopathological features, and clinical outcomes. Larger multicentric studies with longer follow-up periods are required to better

understand the clinicopathological spectrum of pilomatricoma.

7. Conclusion

Pilomatricoma is a benign hair matrix tumor that predominantly affects children and young adults. Although it presents with characteristic histopathological features, clinical diagnosis remains difficult because of its resemblance to other cutaneous lesions. Histopathological examination remains the gold standard for definitive diagnosis. Complete surgical excision is the definitive management. Increased awareness among clinicians and pathologists can facilitate early diagnosis and appropriate management, thereby reducing diagnostic errors and unnecessary interventions.

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