Commonly Misdiagnosed Facial Lesion: Pilomatricoma

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ABSTRACT

Pilomatricoma, also known as Pilamatrixoma or Malherbe's calcifying epithelioma, is a benign skin tumour with a bimodal age distribution between the paediatric and elderly age groups. Although it was previously thought to be rare, recent studies have revealed that it is quite common. Typically, pilomatricoma is diagnosed following histopathological examination of the lesion as it is frequently misdiagnosed with other types of skin pathology. In our case, the child presented with painless swelling of the left infraauricular region. The initial cytology and imaging were unable to provide a definite diagnosis. An excision biopsy was done, and a histopathological examination was suggestive of Pilomatricoma. Therefore, Pilomatricoma ought to be considered in the differential diagnosis of head and neck lesions in hopes of providing a better understanding on this pathological lesion.

KEYWORDS

pilomatrixoma; pilomatricoma; epithelioma of Malherbe; benign mass

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INTRODUCTION

Malherbe and Chenantais, in 1880 described a benign skin lesion which was called the calcifying epithelioma (1). Forbis and Helwig later discovered through histopathological examination that the lesion originated from the matrix cells of hair follicles (2, 3). Since then, this benign lesion was referred to as "Pilomatricoma" or "Pilomatrixoma", which typically manifest in the first or second decade of life. A slight predominance of females was reported. Most of this entity manifested as a single slow-growing, painless, firm mobile lesion. Due to its origin, it adhered to the skin but not the underlying tissue. It is commonly found in the head, neck, and upper extremities, particularly on the face (4) and was typically small, measuring less than 5 cm. Although not uncommon, it was frequently misdiagnosed as other facial lesions such as sebaceous cysts, dermoid cysts, foreign body reaction, calcified lymph nodes, fat necrosis and cartilage (1, 4). It was diagnosed post-operatively by identifying islands of epithelial cells containing basophilic cells, ghost cells, and, on rare occasions, foreign body giant cells and calcifications during histopathological examination (5). The only treatment is surgical resection. It rarely recurs or progresses to cancer. This case report aims to raise awareness of this tumor so that it is not overlooked in the differential diagnosis.

CASE REPORT

A previously healthy 11-year-old boy presented with a two-week history of left infraauricular swelling. He was otherwise well. No other ear, nose or throat symptoms were reported. There were no constitutional or B symptoms (this is the full name. It's a triad of symptoms namely fever, night sweat and significant weight loss which may suggest of lymphoma). No recent sick contact or similar episode in the family. The child's immunization record was up to date, and his developmental milestone was according to age.

On examination, a 1 × 1 cm firm, non-tender, mobile, painless swelling was found in the left infrauricular region. There were no overlying skin changes or palpable neck nodes. Other examinations were normal. He was treated as an infected sebaceous cyst and was given a course of oral antibiotics. However, he defaulted the follow-up due to logistic issue. He returned to us seven months later due to persistent swelling. The lesion remained the same size with no signs of inflammation or infection. Blood infective parameters and tuberculosis screening were within normal parameters.

Ultrasonography demonstrated a well-defined heterogeneous solid lesion in the subcutaneous area of the left infraauricular region measuring 0.7 × 1.3 cm (Fig. 1). Fine needle aspiration (FNAC) of the lesion revealed acellular squames.

The child then underwent excision of the left infraauricular mass. Intraoperatively the lesion was superficial, measuring 1.0 × 1.0 cm, and located beneath the subcutaneous tissue (Fig. 2). The histopathological examination of the lesion showed features suggestive of pilomatricoma. Fig. 3 showed aggregates of foreign body type multinucleated giant cells intermixed with anucleated shadow cells.

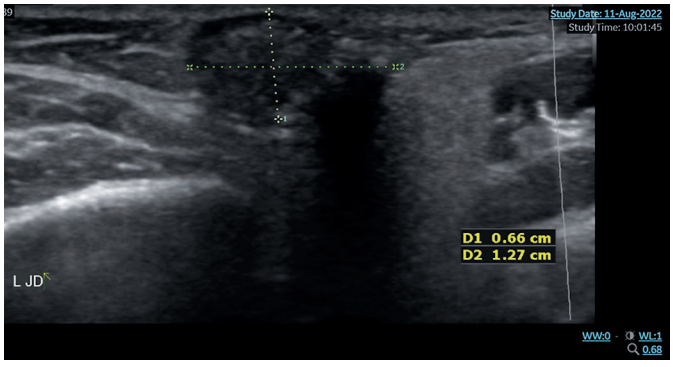

Fig. 1 Ultrasonography of a well-defined heterogeneous solid lesion measuring 0.7 × 1.3 cm.

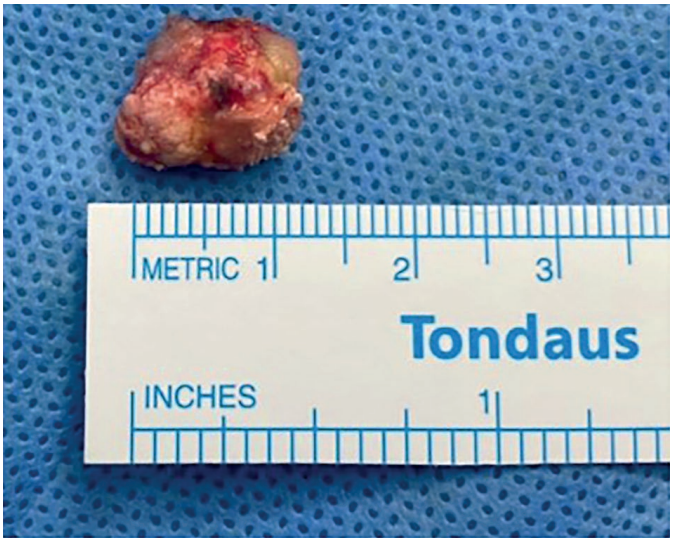

Fig. 2 Histopathological examination of the lesion showing aggregates of foreign body type multinucleated giant cells intermixed with anucleated shadow cells.

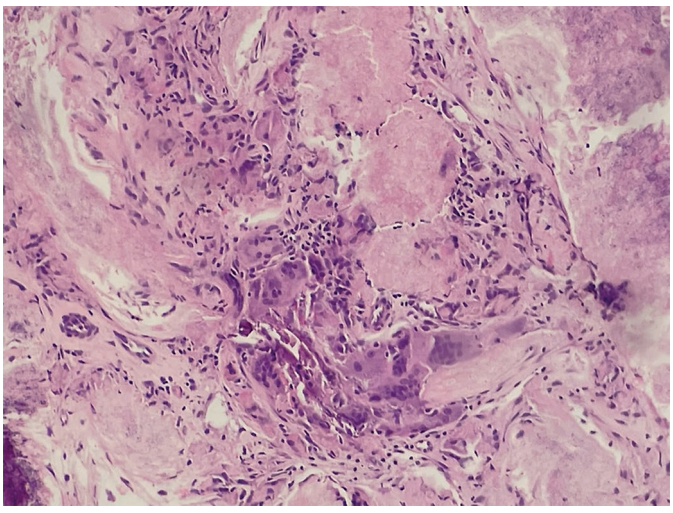

Fig. 3 Nodules of anucleated shadow cells and calcification, with cholesterol cleft.

DISCUSSION

The majority of Pilomatricoma studies revealed bimodal age distribution. It usually peaked in the first two decades or between the ages of 40 to 60 (4, 6). It is commonly found in children. 40% occur before the age of ten, and 20% occur between the age of eleven to twenty (7). It is slightly more common in female (7, 8). The common locations of Pilomatricoma were the head and neck, upper extremities, trunk, and lower extremities (2–4). In addition to that, 40% of them were discovered on the head, particularly in the cheek or palpebral area (4, 7). Nonetheless, despite its frequency, preoperative diagnosis was only achieved in approximately 28.9 to 43 percent of cases due to non-specific presentation and imaging (4).

The most common presentation of Pilomatricoma was solitary, firm, painless swelling, as noted in our case. The lesion commonly located in the deep dermis or subcutaneous layer (3, 4). This gave them the characteristic of skin adherence but mobile from the below structure (1). Due to the presence of blood vessels in the lesion, some will have bluish-red discoloration (7). "Tent sign" was a unique feature of Pilomatricoma. It was caused by tumoral calcification in the skin caused by excessive tension, resulting in tense skin with multiple facets and angles (3, 4). Although the lesion was usually solitary, it can present as multiple nodules in 2-9 percent of cases (4). Multiple lesions were common in people with genetic diseases (8). The genetic disease that was commonly associated with pilomatricoma were Gardner syndrome, myotonic dystrophy and Turner Synrdome (7). Turner Syndrome and myotonic dystrophy account for 42% of genetic diseases associated with Pilomatricoma (7).

Ultrasonography is the most commonly used method because it is non-invasive, non-radiative, inexpensive, and quick, making it ideal for use in children. In ultrasound, the lesion appeared as well-defined, ovoid, hypoechoic, heterogenous masses with or without posterior shadowing (7). In our case, an ultrasound revealed a well-defined heterogeneous solid lesion measuring 0.7 × 1.3 cm in the subcutaneous area of the infraauricular region. Computed tomography is another imaging option for Pilomatricoma, although it is non-specific (9), with well-defined subcutaneous mass seen with mild to moderate enhancement with calcifications.

The classical cytological triad features of Pilomatricoma include basaloid cells, ghost cells and giant cells, albeit present in only 40% of cytology (7). Moreover, only 44% of the lesions can be correctly diagnosed using FNAC (7) which results in a diagnostic dilemma. In our case, FNAC revealed acellular squames, whereas histopathological examination revealed a triad of basaloid epithelium, ghost cell, and giant cell, indicating Pilomatricoma.

Differential diagnosis for Pilomatricoma include sebaceous cysts, dermoid cysts, foreign body reaction, calcified lymph nodes, fat necrosis and cartilage (1, 4). The majority of them are difficult to distinguish based on history and clinical examination, and will require a histopathological examination.

It is worth noting that, Pilomatricoma does not regress on its own and cannot be treated using pharmacological treatment (6). The primary treatment for it is complete surgical resection. Because the preoperative diagnosis is usually speculative, surgical resection is typically deemed for diagnostic confirmation. Additionally, a low recurrence rate of 0–3% (6) had been documented, usually caused by incomplete resection (7). Recurrence in this entity occur between a year and a decade after resection (4). In view of the low recurrence rate, long-term follow-up is not required. Yet, malignancy must be ruled out in patients suspected of Pilomatricoma recurrence, albeit rare and involving the elderly age group (4). So far, only one case of malignant transformation in children with Pilomatricoma has been reported (4). In the same vein, a safe surgical margin of 1–2 cm is required for the excision of pilomatricoma carcinoma as it has been reported to metastasize. Hence, long-term follow-up is warranted post-resection.

CONCLUSION

Pilomatricoma is a relatively common benign skin tumour in children, which is frequently overlooked. However, by increasing the awareness, more research can be carried out to better understand this entity.

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