

# Pilomatricoma Located at the Identical Position in the Right Upper Limb of the Twins: A Rare Case Report

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**Introduction:** Rare studies have reported pilomatricoma in twins, and extremely rare cases showed lesions in the same part of the body position. We reported a case of monozygotic twins with pilomatricoma in the same location on the skin of the right upper extremity.

**Case Presentation:** Seven-year-old monozygotic twins presented to our department with a palpable, painless, and solid mass in the subcutaneous tissue of the right upper limb. A 1.5-cm diameter nodule was seen on the anterolateral aspect of the right upper extremity of the twins. The node was irregular in shape, and upon palpation, the patients reported no noticeable tenderness. Following the administration of local anesthesia, the twins underwent surgical procedure to excise the solid mass. Finally, they were diagnosed with pilomatricoma based on the clinical and histopathological features. Complete surgical resection followed by primary closure was performed. During a follow-up period of three years, there has been no recurrence observed in the twins.

**Conclusion:** We reported a case of monozygotic twins with pilomatricoma in the same location on the skin of the right upper arm. Our findings underscore the requirement of considering genetic factors in the diagnosis and treatment of the rare conditions.

**Keywords:** pilomatricoma, pilomatrixoma, calcifying epithelioma of malherbe, case report

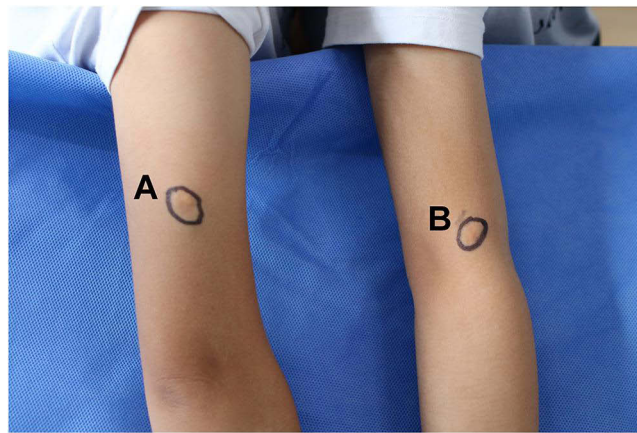
## Introduction

Pilomatricoma, also known as calcifying epithelioma of Malherbe, is a benign cutaneous tumor that originates from the hair matrix cells. Initially described in 1880, the entity was characterized as a calcified tumor derived from the sebaceous gland with clinicopathological characteristics of a firm, asymptomatic, and subcutaneous nodule.<sup>1</sup> The nodule, often found in the head and neck regions, as well as the upper extremities, typically presents in adolescents.<sup>2-4</sup> Less frequently, the nodule has also been observed on the trunk and extremities.<sup>5,6</sup>

Pilomatricoma is more frequently observed in individuals of the white race and shows a predominance in females.<sup>7</sup> To date, the genetic underpinnings of pilomatricoma remain poorly understood, attributed to the condition's relative rarity. Indeed, there are few studies describing the pilomatricoma in twins,<sup>8</sup> while there are no reports of twins developing pilomatricoma at the same body position. In this case report, we present a rare occurrence involving monozygotic twins both diagnosed with pilomatricoma at identical locations on the skin of their right upper extremities. This rare case offers valuable insights into the pathogenesis, diagnosis and treatment of pilomatricoma, potentially advancing our understanding of this uncommon condition.

## Case Presentation

Monozygotic twins, 7 years old, presented to our department with a palpable, painless, and solid mass in the subcutaneous tissue of the upper right limb on August 14, 2019 (Figure 1). This lesion was present at the age of 2 years, and a gradual growth was observed in the last month. The twins had no other medical history. No relevant family



**Figure 1** Pilomatricomas on the right upper arms of twin 1 (A) and twin 2 (B) visualized with irregular nodules (black circles).



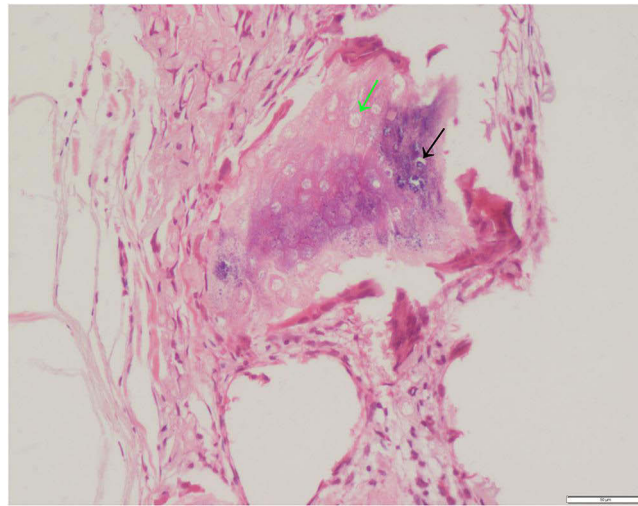
**Figure 2** Intraoperative specimen.

history was found to be a predisposing factor for pilomatricoma. Upon clinical examination, a 1.5-cm diameter nodule was identified on the anterolateral aspect of the right upper extremity in both twins, located approximately 10 cm above the elbow. The nodules were irregular in shape, and no obvious tenderness was reported by the patients on palpation. Following administration of local anesthesia, both twins underwent surgical excision of the solid masses. The mass presented a firm texture and a grayish-white color, with well-circumscribed boundaries (Figure 2). Histological examination revealed the presence of basophilic basaloid cells and ghost cells, confirming the diagnosis of pilomatricoma (Figure 3). During a follow-up period of three years, there was no evidence of recurrence.

## Discussion

Pilomatricoma, a benign epithelial neoplasm originating from hair follicle matrix cells, was first identified as a calcified epithelioma by Malherbe in 1880, named for the calcifications observed within the sebaceous glands.<sup>1</sup> The histological hallmark, characterized by the presence of islands of epithelial cells and shadow cells, was described by Dubreuilh and Cazenave in 1922.<sup>9</sup> In 1961, Forbis and Helwig introduced the term “pilomatricoma” to describe this solitary, slowly growing lesion in the subcutaneous tissue.<sup>10</sup> To date, there are few studies describing the pilomatricoma in twins. In this study, we report on 7-year-old monozygotic twins diagnosed with pilomatricoma located identically on the skin of the right upper extremity, hinting at a potential genetic predisposition towards pilomatricoma.

Pilomatricoma frequently affects the head-neck regions and extremities of the pediatric population. In this case, the twins presented with a single, asymptomatic, solitary, and subcutaneous nodule on the skin of the right upper arm. While



**Figure 3** Photomicrographs showed epithelial islands consisting of basaloid matrix cells and ghost cells (Hematoxylin & eosin; bar scale: 50  $\mu$ m); Basaloid matrix cells (black arrow) exhibiting hyperchromatic nuclei without scant cytoplasm. Ghost cells (green arrow) characterized by pale and eosinophilic appearance, and center empty zone.

the exact etiology of pilomatricoma remains not fully elucidated, studies point out that disruption of the hair follicle cycle, trauma, and infection can lead to the development of pilomatricoma. Considering the consistent phenotypic characteristics of the nodules (in terms of size and localization), the causative factors such as trauma and infection were ruled out. Monozygotic twins share nearly all of their genetic material, so the occurrence of pilomatricoma in both twins could indicate that genetic factors play a significant role in the development of this condition.

The relevant genetic data have revealed the molecular pathophysiology in both malignant pilomatricoma and benign pilomatricoma, which has been elucidated through whole-exome genomic profiling.<sup>11,12</sup> Benign pilomatricoma harbors the classical *CTNIB1* (the gene encoding catenin beta 1 protein) mutant.<sup>11</sup> Zhang et al reported that pGlu396Lys variant of *PLCD1* from germline causes gain-of-function of PLC $\delta$ 1, which is a causative genetic defect in familial multiple pilomatricoma.<sup>12</sup> Additionally, mutations in *CREBBP* and *EP300* have been described in patients with pilomatricoma and Rubinstein-Taybi syndrome.<sup>8,13</sup> Thus, we speculated that gene mutation of the monozygotic twins may be associated with the onset of the solitary lesions. However, research into cutaneous conditions often reveals complex interactions between multiple genes and environmental factors, rather than single-gene mutations. Thus, this conjectural mechanism warrants further investigation, such as whole-exome sequencing, to reinforce the link between pilomatricoma and genetic mutation.

The diagnosis of pilomatricoma relies exclusively on histopathological findings, which include the presence of anucleate pink ghost cells and aggregates of small round blue cells, representing a matrical epithelial component akin to the round blue cells found in the hair bulb of normal hair follicles.<sup>14–16</sup> The nodule is generally surrounded by brisk granulomatous inflammation, fibrosis, and foreign body giant cell reaction to the keratin from the anucleate ghost cells.<sup>17</sup> Calcifications usually appear and metaplastic bone may form.<sup>18,19</sup> Mitoses may be detected in the round blue cell matrical component, which may not support malignant onset.<sup>20</sup> In our study, histological examination of the twins suggested the presence of epithelial islands composed basaloid matrix cells and ghost cells, which are the hallmark features of pilomatricoma.

In clinical practice, despite its benign nature, the rarity of the condition, especially in twins, can lead to a wide range of differential diagnoses before a definitive diagnosis is made. Pilomatricoma should be differentially diagnosed from dermoid or epidermoid cysts, dermoid cysts, foreign body granuloma, basal cell carcinoma, calcified lymphadenopathy, ossified or calcified hematomas, giant-cell tumor, lipoma, and trichoblastoma.<sup>5,21</sup> Ultrasonography and computed tomography are frequently employed in the differential diagnosis of pilomatricoma, suggestive of heterogeneity, well-defined ovoid margin, hyperechogenicity or isoechoic heterogeneity, peripheral hypoechoic rim, posterior acoustic shadowing, well-marginated subcutaneous mass, adherent to the overlying skin, and variable amounts of

calcification.<sup>22</sup> Epidermoid cysts, the most prevalent type of cutaneous cysts, can be challenging to distinguish from pilomatricoma, which showed a round to oval structure, well-circumscribed, vascular mass located in subcutaneous tissue along with phenomena of dorsal acoustic amplification and lateral shadowing in the ultrasound images.<sup>23</sup> Cutaneous hemangiomas were ruled out considering the propensity to infants and the characterization of abundant colored blood flow.

## Conclusions

We reported twins showing the typical presentation of pilomatricoma in the same location on the skin of the right upper arm, which is extremely rare. The diagnosis of pilomatricoma was based on the histopathological features that present anucleate pink ghost cells and aggregation of small round blue cells. Complete surgical resection with primary closure is the preferred treatment. There has been no recurrence in our case during 3-year follow-up. Our findings strongly suggest a genetic component to pilomatricoma in monozygotic twins. This underscores the importance of considering genetic factors in the diagnosis and treatment of rare conditions, especially in twins, and highlights the need for a multidisciplinary approach that includes genetic counseling and psychological support for affected individuals and their families.

## Data Sharing Statement

The datasets generated during and analyzed during the current study are available from the corresponding author on reasonable request.

## Ethical Approval

The research related to human use has been complied with all the relevant national regulations, institutional policies and in accordance the tenets of the Helsinki Declaration, and has been approved by the Ethical Committee of The Second Hospital of Jilin University (approval No.: 2022-094). Informed consent has been obtained from the patient's parents included in this study.

## Consent for Publication

Written informed consent for publication was obtained from the parents of the patient.

## Author Contributions

All authors made a significant contribution to the work reported, whether that is in the conception, study design, execution, acquisition of data, analysis and interpretation, or in all these areas; took part in drafting, revising or critically reviewing the article; gave final approval of the version to be published; have agreed on the journal to which the article has been submitted; and agree to be accountable for all aspects of the work.

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## Disclosure

The authors declare that they have no competing interests in this work.

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