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Case Report and Highlight Clues on the Diagnosis of Pilomatrical Carcinoma

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Abstract

Pilomatrical Carcinoma (PC) is a rare malignant adnexal tumor with matrical differentiation. Its benign counterpart (Pilomatrixoma) is diagnosed much more frequently in daily pathological practice. Both entities share genetic alterations but the malignant counterpart acquires mutations that make it develop an aggressive behavior [1].

We describe a 33-year-old man who presented with a 7 x 6 cm nodular ulcerated lesion in the left ear with markedly accelerated growth in the last month. Incisional biopsy was referred to us with suspicion of squamous cell carcinoma versus pyogenic granuloma.

Histologic sections showed ulcerated fragments infiltrated by a basaloid cell proliferation interspersed with groups of "ghost cells". The neoplastic cells were arranged in irregular sheets with infiltrative borders. Groups of Squamous cells with trichilemmal keratinization and foci of necrosis were also identified. The biopsy was diagnosed as an adnexal neoplasm with pilomatrical differentiation, suggesting its complete resection with safety margins due to the presence of aggressive characteristics. The subsequent study of the excisional biopsy showed similar characteristics to those previously described. Notoriously, focal infiltration of the auricular cartilage was identified, leading us to the undoubted diagnosis of pilomatrical carcinoma.

Keywords

Case Report, Pilomatrical Carcinoma, Cutaneous Adnexal Tumors, Pilomatrixoma

Introduction

Pilomatrical carcinoma (PC) is a malignant adnexal neoplasm with matrical differentiation that shows a very low incidence. To this date, approximately about 150 cases reported in the literature so far [2]. There are no risk factors for these tumors. However, as they frequently occur in sun-exposed areas such as the head and neck (60%), previous studies suggest that UV radiation may contribute as an oncogenic driver to malignant transformation [3,4].

In addition and also reported in the literature, PC and pilomatrixoma are known to be related and share similar genetic alterations but PC probably acquires additional mutations over time that endow it with more aggressive behavior [1].

Regarding molecular biology, *CTNNB1* (gene encoding β-catenin) is the only driver mutation identified and accepted in both benign and malignant tumors. Nowadays a distinct and novel tumorigenic pathway which involves fibroblast growth factor receptor 4 (FGFR4) mutation is being studied [2].

Clinically and epidemiologically, in contrast to pilomatrixoma, PC usually occurs more frequently in men during the fifth to seventh decade of life, as a large tumor (up to 10 cm) with poorly defined margins and with an ulcerated surface.

Due to its slow growth and rarity, it is often clinically misdiagnosed as a sebaceous cyst or other common skin tumor, potentially delaying tumor resection [5]. Limited biopsy may miss key histopathological features because of limited tissue sampling and make the diagnosis considerably challenging [6].

Because of the malignant nature of the lesion, surgical treatment it is usually performed. Adequate margin resection measurement is controversial. Margins between 5mm-2cm are reported as sufficient. In some cases, Mohs surgery has been described as an alternative approach [3,7].

As informed in the literature, 23% of patients may develop local recurrence within the first 7 months despite the presence of clear surgical margins [3]. Metastatic disease is extremely rare, being lymph nodes and lungs the most frequent location [1,8].

What makes this case unique, apart from the fact that PC is an exceptional tumor, is the fact that due to a detailed microscopic observation of a very small biopsy, we were able to identify the little and few characteristic clues of malignancy. This allowed us to make a wise warning to the surgeon and guide the behavior towards a correct management of the patient.

Presentation of the Case

We describe a 33-year-old male who was admitted to our hospital with non-previous medical condition, consulting for a tumoral lesion noticed since an indeterminate time ago. It remained stable and invariable for several months but unexpectedly an accelerated an exponential growth occurred during the last month. The patient reported moderate pain and was concerned about the rapid growth and ulceration of the lesion.

On physical examination, a 7 x 6 cm brownish ulcerated firmly attached nodular tumor was recognized. It compromised almost the entire surface of the left outer ear and showed poorly defined edges (**Fig-1**). No palpable lymphadenopathy was observed in the region or the surrounding area. The rest of the physical examination was normal with stable vital signs. An incisional biopsy was referred to our service for histopathological study with clinical suspicion of squamous cell carcinoma versus pyogenic granuloma.

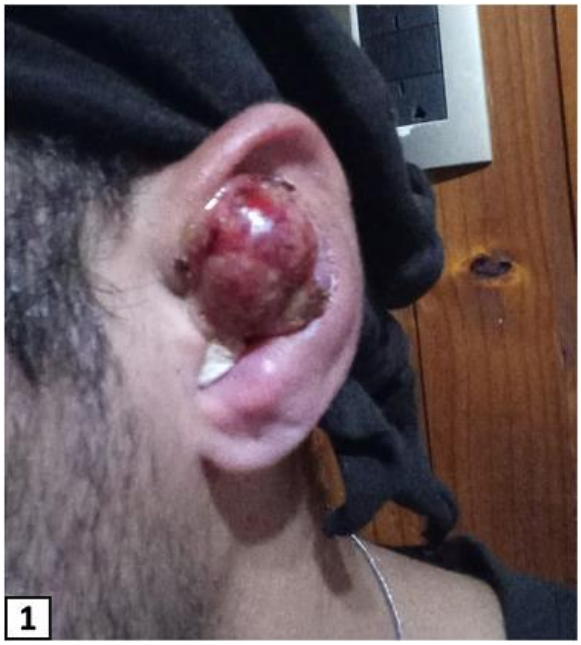

Fig-1: Clinical image of the ulcerated nodular tumor in the outer left ear

The microscopic study revealed an ulcerated fragment covered by fibrinoid material and abundant granulation tissue (**Fig-2**) infiltrated by a proliferation of basaloid cells with scant cytoplasm and hyperchromatic, round to oval nuclei (**Fig-3**). The cells were arranged in irregular sheets, nests and broad cords with an infiltrative growth pattern (**Fig-4**). They alternated with groups of "ghost cells" with extensive eosinophilic cytoplasm, surrounded by foreign body reaction with giant cells (**Fig-5**), and squamous cells with trichilemmal keratinization. Additionally, osseous

metaplasia (**Fig-6**) and foci of necrosis and suppuration were identified (**Fig-7**).

The lesion was diagnosed as an adnexal neoplasm with pilomatrical differentiation, suggesting a complete resection with safety margins due to the presence of aggressive characteristics such as necrosis, infiltrative border, size and ulceration. Immunohistochemistry was not necessary for the diagnosis, as we have previously mentioned, it is not useful nor decisive for the diagnosis.

A few days later, complete resection of the lesion was performed with margins of between 1 and 0.5 cm. The team of plastic surgery surgeons was able to reconstruct the pinna.

The excisional specimen study showed similar characteristics to those previously described and in addition we could certify focal infiltration of the cartilage (**Fig-8** and **Fig-9**). These findings reaffirmed the diagnosis of PC. In the extensive sampling and total inclusion of the material, we did not find sectors that were clear and consistent with pilomatrixoma remains.

The patient recovered from the surgery without complications. The healing and reconstruction of the tissue evolved correctly with the help of plastic surgery. Complementary studies were carried out in search of lymph node metastases and distant metastases, which were negative. Currently, 4 months have passed since the surgery and in the follow-up of the patient there have been no recurrences and no other relevant findings.

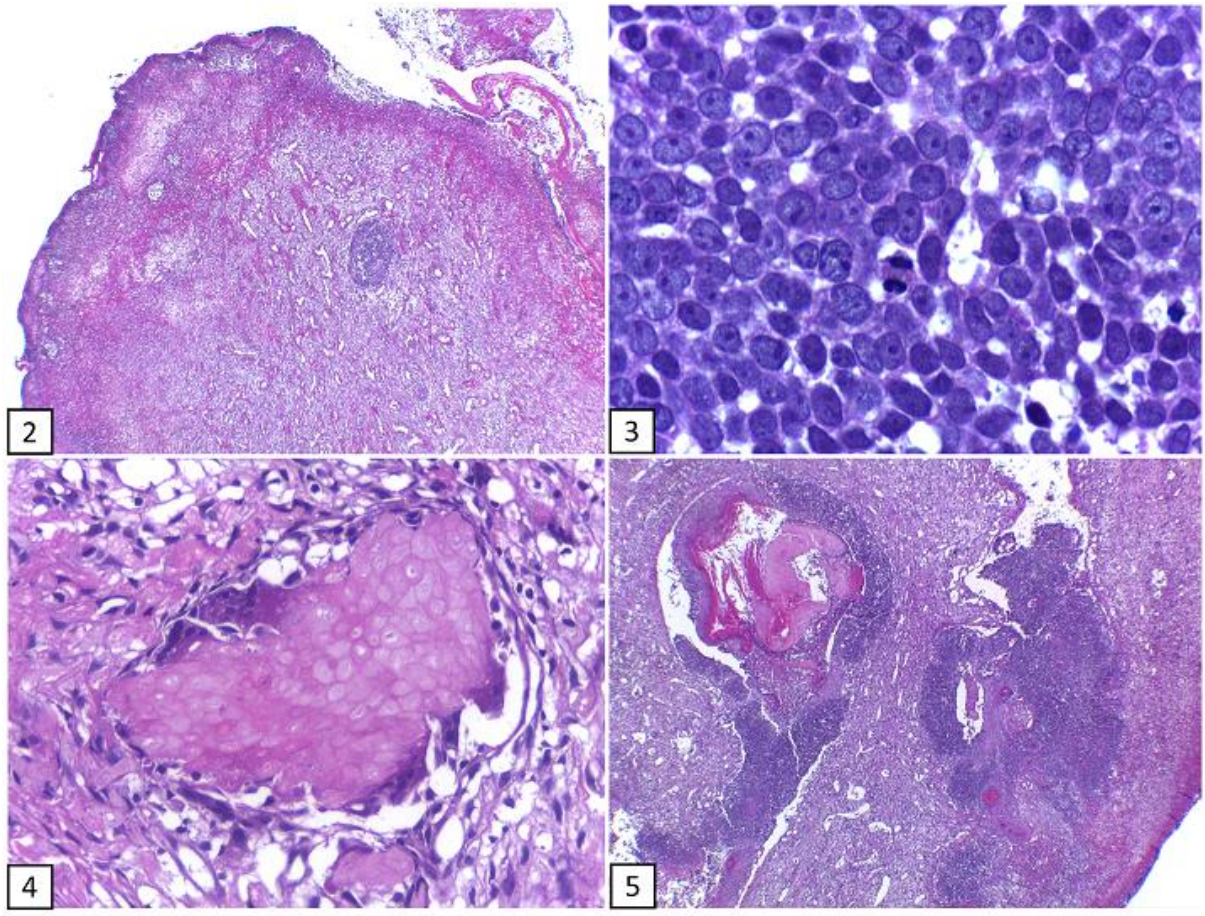

Fig-2: H&E 40x: Ulcerated fragment covered by fibrinoid material and abundant granulation tissue
Fig-3: H&E 1000x oil: Proliferation of basaloid cells with scant cytoplasm and hyperchromatic, round or oval nuclei
Fig-4: H&E 40x: Proliferation arranged in nests and broad cords, with infiltrative-like growth margins
Fig-5: H&E 100x: Groups of "ghost cells" with extensive eosinophilic cytoplasm, surrounded by a foreign body reaction

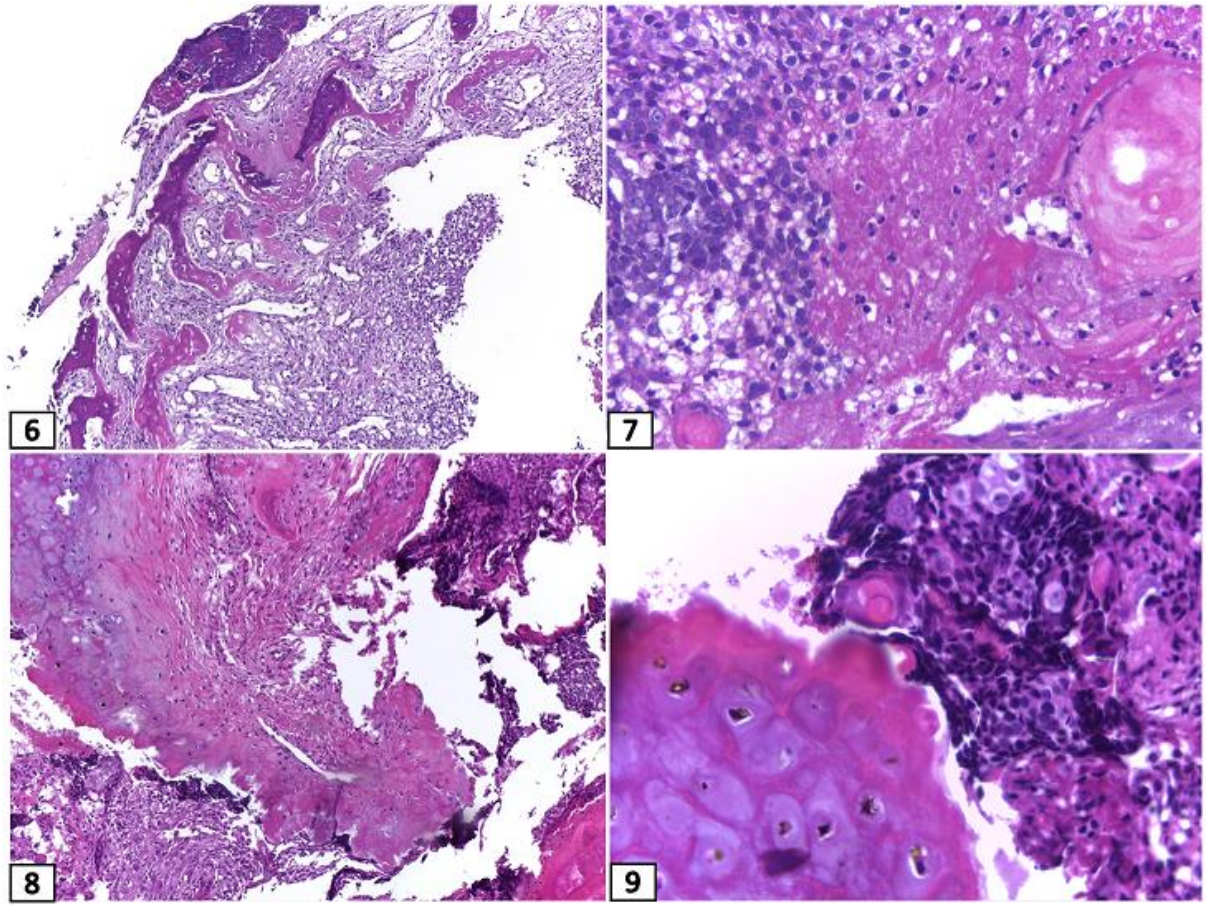

Fig-6: H&E 40x: Osseous metaplasia
Fig-7: H&E 400x: Foci of necrosis
Fig-8: H&E 40x: Basaloid neoplasm infiltrating cartilague
Fig-9: H&E 1000x oil: Basaloid neoplasm infiltrating cartilague

Conclusion

PC is a rare entity with few cases reported in the literature. It is considered a low-grade neoplasm, with substantial rates of local recurrence but low frequency of metastases, mainly to lymph nodes and lungs [8] and it is clinically often misdiagnosed. Taking this data into account, it was important for us to be familiar with this entity, to highlight the chronology of our patient´s study and the reasons why we arrived to the final diagnosis. These may be useful for the study of future lesions of similar characteristics.

Clinical and histological features of PC are diverse and simulate other tumours (pilomatrixoma, basal cell carcinoma with matrical differentiation, etc.), that can lead to diagnostic errors. It is appropriate to seek the clinical and pathological characteristics of aggressiveness. Macroscopic and microscopic features suggestive of malignancy include size greater than 4 cm, necrosis, infiltrative borders, predominance of basaloid cells, atypical mitoses, nuclear atypia, compromise of deep tissue, vasculolymphatic and perineural invasion [5]. The immunohistochemical profile has been studied to differentiate benign and malignant counterparts but was not useful to make this distinction.

It is important to spotlight certain situations in particular, as happened in our case, were in first instance we only received a small and incisional biopsy. In such cases it might not be possible to find all malignancy characteristics but at the slightest suspicion of aggressive behavior it is relevant to report it and suggest a wider resection.

The main strengths of this study were the possibility of alerting the surgeon to our suspicion in the biopsy, with the followed act of an expeditious excision

surgery (in less than 1 month after our diagnosis in the biopsy) and the subsequent certification of malignancy and tumoral infiltration of the auricular cartilage in the free margin resection piece. Tanks to the fact that all these events developed correctly, the patient got cured.

We report three main limitations in our case: the lack of information about the time of growing evolution of the lesion, not having found remnants of Pilomatrixoma and the shorter follow-up. Although the first two is not essential for the diagnosis, they would have helped us. Regarding the follow-up, we will keep in contact with the patient to find out his long-term evolution.

The primary upskill from this case report was learning the importance to be alert of malignant characteristics of PC, even when limited, in order to achieve and guide the best medical practice, treatment and patient follow-up.

Conflict of Interest

The authors have read and approved the final version of the manuscript. The authors have no conflicts of interest to declare.

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