

Case Report

Cylindroma – a case report: benign but not banal!

圓柱瘤病例報告 – 良性卻非凡

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The diagnosis of cylindroma can be challenging due to the rarity of the tumour and its broad differential diagnosis. We present a case report of a solitary benign cylindroma on the right scalp of a 51-year-old patient which was initially thought to be a basaloid follicular hamartoma based on biopsy. We outline the differential diagnosis of cylindroma, as well as the histopathological features and the pathological differences between cylindroma and basaloid follicular hamartoma. The ethical dilemmas surrounding the management of benign solitary cylindromas, including the potential for malignant transformation, risks associated with operating, and cost for the patient, are also discussed.

由於圓柱瘤的罕見及其廣泛的鑑別診斷，其診斷實具挑戰性。我們報告一名51歲患者的右側頭皮上長出單發性良性圓柱瘤的病例報告，該病例最初基於活組織檢查被誤認為基底細胞樣毛囊錯構瘤。我們概述了圓柱瘤的鑑別診斷、組織病理學特徵，以及它與基底細胞樣毛囊錯構瘤之間的病理學差異。並圍繞著良性孤立性圓柱瘤治理上的倫理困境討論，包括惡性轉化的可能性、手術相關的風險以及患者的付出代價等等。

Keywords: Adnexal skin tumour, basaloid follicular hamartoma, cylindroma, linear and unilateral, malignant transformation

關鍵詞： 附件皮膚腫瘤、基底細胞樣毛囊錯構瘤、圓柱瘤、線性和單側、惡變

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Introduction

Cylindromas are rare adnexal skin tumours arising from eccrine or apocrine glands. Cylindromas typically occur on the head and neck and predominantly affect women, with the male to female ratio ranging from 1:3 to 1:9.¹ They can occur as solitary lesions, or as multiple lesions, which can be associated with familial cylindromatosis, inherited in an autosomal dominant form, or Brook-Spiegler syndrome.

Genetic mutations in the cylindromatosis tumour suppressor gene (CYLD1) on chromosome 16q12-13 have a role in the development of both hereditary and sporadic cylindroma.^{2,3} Malignant cylindroma is a rare tumour arising from pre-existing benign cylindroma, and can occur in both the solitary and multiple types, although malignant transformation occurs more commonly in the multiple type.^{4,5}

Case report

A 51-year-old woman presented for review of a row of slowly enlarging lesions on her right scalp and forehead (Figures 1 & 2). She had no significant past medical history, took no regular medications and reported an allergy to penicillin. She had no family history of similar skin lesions and her two children were unaffected. The lesions, which were asymptomatic, had been present for fourteen years. It is of interest to note that the lesions are unilateral, clustering on the right side of the scalp. One of the larger lesions on her scalp was excised by a general surgeon in 2008 and the biopsy report favoured a diagnosis of cylindroma. However, a subsequent and relatively

superficial biopsy in 2012 was considered to be a basaloid follicular hamartoma. For clarification, further biopsies were taken and these showed features of cylindroma (Figures 3 & 4).

Discussion

The differential diagnosis to be considered in cylindroma includes: basal cell carcinoma;⁶ trichilemmoma; trichoepithelioma; basaloid follicular hamartoma; spiradenoma and neurofibromatosis. Cylindromas may also be mistaken for a keloid scar. As can be seen in the photos below, a cylindroma may closely resemble a nodular basal cell carcinoma. Moreover, an amelanotic melanoma or a Merkel cell carcinoma can also present in a similar way. Regular follow-up is important to ensure that skin cancers are excluded and treated appropriately if any further lesions develop.

The distinction between cylindroma and basaloid follicular hamartoma is usually fairly straightforward in excision specimens, but may be difficult in small and superficial biopsies.



Figures 1 and 2. These figures illustrate the linear and vertical lesions on the patient's right scalp. The largest lesion at the top of the photo resembles a nodular basal cell carcinoma.

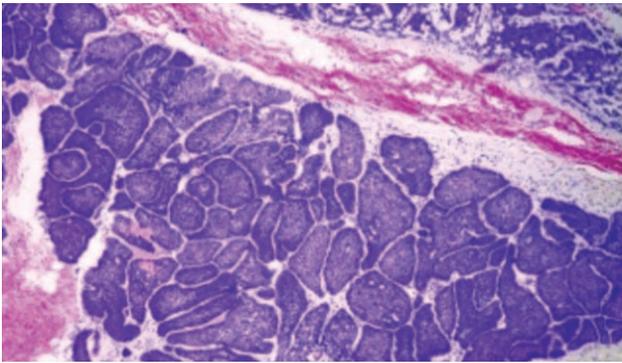


Figure 3. Typical cylindroma, with a 'jigsaw' like histology of islands of basaloid cells with surrounding hyaline thickened basement membrane material.

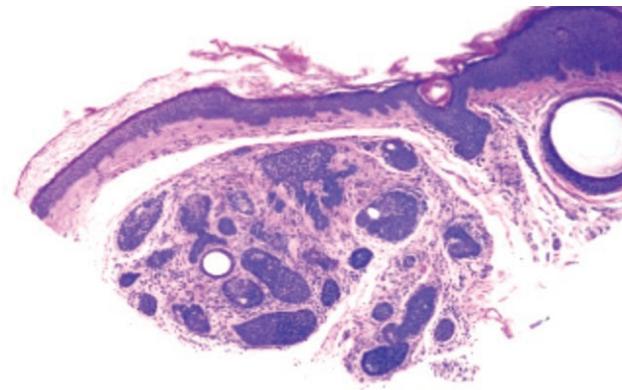


Figure 4. The patient's last biopsy showing small islands of basaloid cells, some with the hyaline material, but others with cellular stroma reminiscent of perifollicular mesenchyme.

Basaloid follicular hamartoma has two distinct histological appearances. Both show a dermal based tumour without connection to the epidermis. The first pattern is a collection of small islands with a somewhat net-like architecture in the superficial to mid dermis associated with pilosebaceous units at the infundibular level. These islands are composed of peripherally palisading basaloid cells and some central squamous cells. There is a cellular surrounding stromal component, composed of bland spindled cells with some abortive hair bulb like structures, and CD34 positive cells. The second histology is that which is very reminiscent of trichoepithelioma, with larger

expansile anastomosing lobules and trabeculae of similar cells, and with similar surrounding stromal changes to those described above.

Cylindroma is a dermal based tumour, also without connection to the epidermis, composed of multiple lobules arranged in a 'jigsaw' like architecture, with surrounding thick DPAS positive hyaline basement membranes. The lobules are composed of two cell types: small basaloid cells and scattered larger cells with pale cytoplasm. Some ductal lumina are usually present. Occasionally the lobules also contain hyaline droplets. Cylindroma, composed of lobules of basaloid cells, is, in these respects, very similar to basaloid follicular hamartoma. However, cylindroma usually demonstrates a hyaline, DPAS positive thickened basement membrane, as opposed to the cellular fibrous stroma of basaloid follicular hamartoma. The cells of both tumours may be predominantly basaloid, although in cylindroma, there is often the admixed larger myoepithelial type cells, together with hyaline droplets and occasional ductal lumina. The basaloid tumour cells in cylindroma show positive staining for CEA and EMA, which is not seen in basaloid follicular hamartoma. The larger cells in cylindroma show positive staining for SMA and S100 protein, which is also not seen in basaloid follicular hamartoma.

The question of how to manage a solitary benign cylindroma is a difficult one given high recurrence rates and the very rare potential for malignant transformation. The clinical features of malignant lesions include rapid growth, ulceration and bleeding.⁵ The prevalence of malignant cylindroma is not well-established due to the rarity of the lesions. Of a total of 36 cases of malignant cylindroma documented in the literature, only nine of these transformed from the solitary type.⁴ Metastatic spread has been reported in 11 patients with lymph nodes, stomach, thyroid, liver, lung and bones affected.⁷ There are several management options for solitary benign cylindroma documented in the existing literature,

including wide local excision, Mohs' micrographic surgery and laser ablation.⁴ Multiple cylindromas and 'turban tumours' which cover the whole scalp typically require extensive surgery with skin grafts.¹ Wide excision is generally the preferred treatment option for solitary cylindroma due to the malignant potential of the tumour, and also for the more accurate histopathological diagnosis obtained from examining an excision specimen. However, it is known that factors such as incomplete surgical removal, trauma, radiation and chronic irritation can contribute to malignant transformation.⁴ The clinician must also consider putting the patient through the risks of the operation including the risk of the anaesthetic, scars and potential disfigurement, with only a very rare documented risk of malignant transformation.

Conclusion

Cylindromas are important clinically, histologically and cosmetically. They are also a sentinel example of dermatologically induced disfigurement. Optimal management is not always straightforward since factors such as ethical and

medico-legal need thoughtful consideration. Our patient is currently well and considering surgical options.

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