CASE REPORT

Proliferating trichilemmal tumour: diagnostic challenge on core biopsy

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SUMMARY

A 70-year-old woman presented with a 10-month history of an irregular mass in the left lateral nape of her neck which had recently increased in size rapidly. Ultrasound-guided core needle biopsy was obtained, and the tumour was diagnosed as a well-differentiated squamous cell carcinoma. Further imaging studies failed to demonstrate additional malignant characteristics. In view of these findings, a wide local excision of the tumour was performed. Histopathological assessment of the resected tumour revealed a proliferating trichilemmal tumour with well-differentiated features and smooth invasion front. This article serves as an important reminder of the challenges associated with pathological evaluation of core needle biopsies of adnexal tumours. It emphasises the importance of clinical-radiological-pathological correlation preferably in a multidisciplinary team setting prior to agreeing on a definitive management plan.

BACKGROUND

A proliferating trichilemmal tumour (PTT) is a well-circumscribed subcutaneous lesion with squamoid cytological features and trichilemmal-type keratinisation.¹ Other diagnostic acronyms include proliferating trichilemmal cyst, proliferating epidermoid cyst, pilar tumour of the scalp, giant hair matrix tumour, hydatidiform keratinous cyst, trichochlamydoid carcinoma and invasive hair matrix tumour.² PTTs have an incidence of 5%–10%, a strong female predominance and age of presentation spanning the fourth to eighth decades. PTTs present typically on the posterior scalp as a slow but progressively enlarging lesion resulting in an exophytic mass.^{3–5}

CASE PRESENTATION

A 70-year-old woman was referred to the Head and Neck Unit at University College London Hospitals, NHS Foundation Trust (UCLH) with an irregular lump in the left lateral nape of her neck first noticed 9–10 months ago with recent rapid increase in size. Her medical history is significant for only chronic obstructive pulmonary disease and hypothyroidism managed with Ventolin and thyroxine, respectively. Physical examination showed an irregular, non-tender 4×5 cm mobile mass not evidently fixed to the skin. No associated lymphadenopathy was identified.

INVESTIGATIONS

Ultrasound and core needle biopsy

Ultrasound imaging revealed an exophytic lesion in the left paramidline of the posterior neck confined

within the subcutaneous tissues in its deep aspects, with no evidence of infiltration into the deep extensor musculature (figure 1A). Internal vascularity was noted within the lesion (figure 1A). Ultrasound-guided core needle biopsy (CNB) was reported as fibrous connective tissue infiltrated by islands of well differentiated squamous cell carcinoma (figure 1B). In view of the unusual location of this lesion, the differential diagnosis included an adnexal tumour as well as an unknown primary squamous cell carcinoma. If the former was not in the differential diagnosis, the management would have involved radical surgery and possibly postoperative radiotherapy to the tumour bed.

MRI and positron emission tomography CT imaging

Further investigations with MRI of the head and neck revealed a well-defined superficial subcutaneous mass in the left paramidline of the neck (figure 1C) within which were scattered areas of mineralisation (figure 1D). No peripheral soft-tissue enhancements and no infiltration into surrounding soft tissues were seen, indicating a non-aggressive tumour (figure 1C,D). Hybrid FDG (fluorodeoxyglucose)-positron emission tomography (FDG-PET) and CT showed no evidence of any primary malignancy throughout the body but it revealed mildly hypermetabolic bilateral upper posterior cervical lymph nodes with the left more prominent than the right. However, this finding was thought to be non-specific. Considering these findings, a primary tumour on the scalp was thought to be the most likely diagnosis.

Therefore the multidisciplinary team (MDT) agreed a surgical resection of the tumour and the localised FDG-avid lymph nodes via a wide local skin and subcutaneous excision in the first instance. However, if on excision the tumour revealed obvious histopathological features of malignancy and metastasis within the excised lymph nodes, the initial management plan, that is, bilateral posterior neck dissection and postoperative radiotherapy to the tumour bed, would be carried out.

DIFFERENTIAL DIAGNOSIS

The core biopsy differential diagnosis will depend on the following features:

- A. Minimal cytological and architectural atypia and lack of stromal invasion:
 - Benign PTT.
 - Well differentiated squamous cell carcinoma.

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Figure 1 (A) Ultrasound image shows no infiltration into deep muscles (white arrows) and the tumour vascularity. (B) Core biopsy histology showing fibrous connective tissue infiltrated by islands of well-differentiated squamous cell carcinoma. (C and D) MRI images of head and neck showing a well-defined lesion in left lateral nape of neck with scattered mineralisation but no obvious features of malignancy (red arrows).

- B. Cytological and architectural atypia, stromal invasion, necrosis, high mitotic rate and abnormal mitoses:
- PTT with aggressive behaviour.
 - Malignant PTT.
 - Moderate to poorly differentiated squamous cell carcinoma.

TREATMENT

Histopathology

The wide local en bloc excision of the skin and subcutaneous tissue revealed a well-circumscribed and lobulated neoplasm with a smooth invasion front in the subcutaneous tissues without overlying skin involvement (figure 2A,B). The tumour was composed of variably sized but mostly large keratinising cystic epithelial islands showing trichilemmal keratinisation and central dystrophic calcifications (figure 2C,D). Budding to form small epithelial nests with squamous eddies was also seen with

Figure 2 Histology images show (A) completely excised tumour with a well-circumscribed and lobulated architecture displaying both a smooth invasion front and clear surgical excision margins, (B) no involvement of overlying skin, (C and D) cystic epithelial islands showing trichilemmal keratinisation and central dystrophic calcification, and epithelial nests with squamous eddies.

the tumour mass (figure 2C). There was no significant cytological atypia, perineural invasion or lymphovascular invasion. Furthermore, the mildly hypermetabolic bilateral upper posterior cervical lymph nodes seen on imaging showed reactive features and no evidence of tumour metastasis histologically. A final diagnosis of a completely excised PTT was made.

OUTCOME AND FOLLOW-UP

Based on the final diagnosis, the resection margins and the absence of tumour metastasis in the excised posterior neck lymph nodes, there was no need for further therapy, such as bilateral neck dissection and postoperative radiotherapy; however, periodic follow-up to assess for recurrence was advised. To date, no local recurrence or tumour metastasis to regional lymph nodes have been reported.

DISCUSSION

PTT is presumed to arise from trichilemmal cysts, and may progress to a malignant PTT. Both tumours can closely mimic squamous cell carcinoma.^{1–5} Therefore, correlation with histopathology is critical to obtain the best patient outcome, and to determine if further adjuvant therapy is required.⁶ Additionally, the similarities in histopathological features of these entities can make evaluation challenging for the reporting pathologist. In this article, we have shown for the first time, the difficulties associated with interpreting CNBs of PTTs without clinical-imaging correlation. This experience highlights the need for caution when reporting CNB from lesions with similar clinical presentation.

The importance of correlation of clinical-imaging-pathological features particularly in an MDT setting cannot be overemphasised as this process ensured the patient received care tailored specifically to their scalp lesion type and size. In other words, the lack of obvious features of malignancy in the imaging studies performed after a CNB diagnosis of well-differentiated squamous carcinoma resulted in a conservative management plan initially. The suitability of this preliminary plan was confirmed in the histopathology of the excised tumour revealing clear excision margins, smooth invasion front and lack of cytological atypia. There was no metastatic tumour in lymph nodes and in the adjacent soft tissue.

PTTs without atypical pathological features are known to behave in a benign manner, however those with an invasive growth pattern and cytological atypia have an unpredictable prognosis.¹ Therefore, long-term follow-up is imperative to mitigate the risk of recurrence and malignant transformation which can occur even after complete excision.^{1,6} Furthermore, by understanding the differential diagnosis for an adnexal tumour, the risk of recurrence/metastasis and long term prognosis can be estimated thereby aiding both the patient and physician in developing a treatment plan.

While the current tumour was a PTT, the extent of disease and tumour type will impact management. For example, in the case of a malignant PTT, surgical excision with a 1 cm margin of normal tissue is the mainstay of treatment.¹ However, if the lesion is in a cosmetically important area, then Moh's micrographic surgery proposed by Fielege *et al* may be considered to limit excision of healthy tissue.⁷ Where local spread of the primary lesion/recurrence suggest more advanced disease, a systemic diagnostic work-up is necessary, and could involve anatomical imaging from radiology (ultrasound and CT), as well as functional imaging from nuclear

medicine (FDG-PET). Neoadjuvant radiotherapy has been proposed both in the medically fragile, and in cosmetically important regions to shrink the tumour before definitive surgical resection.^{7 8} Similar to advanced squamous cell carcinoma, chemotherapy is a consideration in those cases where distant metastases are found, that said, has been reported to have limited success.⁷ Treatment decisions outside of usual therapy are best discussed in an MDT setting and on a case-by-case basis.

Learning points

- ▶ The histopathological features of proliferating trichilemmal tumour (PTT) can closely mimic those of a malignant proliferating trichilemmal tumour, or a well-differentiated squamous cell carcinoma, especially in a core needle biopsy.
- ▶ Caution should be exercised when evaluating and reporting core needle biopsies in cases with similar clinical presentation.
- ▶ Core biopsies should not be used alone to establish a diagnosis, but should be incorporated into a trimodal assessment involving clinical evaluation, hybrid functional imaging and histology.
- ▶ Continued long-term surveillance should form part of the management plan for an excised proliferating trichilemmal tumour.

Contributors NK was involved in the surgical management of the patient and contributed to drafting the case report. SM was involved in the interpretation of the imaging investigations (MRI and CT) and drafting of the case report. OKA and AJ were both involved in pathological assessment of the lesion, establishing a definitive diagnosis, coordinating and preparation of the final manuscript draft. NK, SM and AJ gave final approval for submission of the draft case report.

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