Pilomatrixoma: A Diagnostic Pitfall on Fine-Needle Aspiration Cytology of Benign ‘Neck Metastases’

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ABSTRACT

Pilomatrixoma is a benign tumour that originates from the matrix of the hair root. This rare tumour is usually managed by the dermatologists. The commonest location of this tumour is in the head and neck region; hence, it can be encountered by any doctors with interest in this area. When presented in the neck, this hard tumour may pose a diagnostic challenge. A case report of pilomatrixoma misdiagnosed as a metastatic neck disease from fine-needle aspiration cytology is presented. The mistake in the diagnosis has led to a more aggressive and high morbidity surgery than necessary. It is important that head and neck doctors be aware of this condition and includes it in the differential diagnosis of hard masses presenting in the neck.

KEYWORDS: Pilomatrixoma, benign, head and neck cancer, diagnostic pitfall

INTRODUCTION

Pilomatrixoma is a benign, calcified tumour originating from the outer sheath cell of the hair follicle root. Pilomatrixomas are commonly known to dermatologists and experienced pathologists. This condition is commonly seen in the head and neck region, predominantly in the maxillofacial area. Pilomatrixoma in the neck region can pose a diagnostic confusion. The hard nature of the calcified tumour can be mistaken for a carcinoma in the neck. The diagnosis of pilomatrixoma is fundamentally clinical; however, complementary studies are often needed to establish a differential diagnosis with other lesions in the neck. The complementary studies can lead to a diagnostic pitfall in case of pilomatrixoma.

We present a case of pilomatrixoma misdiagnosed as a malignant cancer from cytology and radiological imaging. This paper serves as a reminder to avoid mistakes in the diagnosis when dealing with the case of hard masses in the neck.

CASE REPORT

A 48-year-old Malay man presented with painless right neck swelling of two-month duration. It was progressively increasing in size. There was no history of fever or other constitutional symptoms. His brother was diagnosed with nasopharyngeal carcinoma a year earlier, after presented with a neck node. He has no other ear, nose and throat complaint. Physical examination revealed a 2 cm rounded hard mass located at the right posterior triangle of the neck (Figure 1). The mass was immobile, and fixed to the skin. Endoscopy of the nose, pharynx, larynx and direct examination in the oral cavity showed no abnormality. With the background family history, the surgeon ordered for fine needle aspiration cytology (FNAC) to rule out metastatic cancer in the neck, with nasopharyngeal cancer (NPC) high on the list of differential diagnosis. The FNAC was reported as metastatic carcinoma (Figure 2A). With this finding, the patient was worked-up for the search of the primary lesion. A thorough endoscopic examination in the upper aerodigestive tract revealed no primary lesion responsible for this possible nodal metastasis. Blood investigation for Eppstein-Barr virus titre and NPC serology was normal. Computerised tomography (CT) scan of the skull base until the thorax showed no abnormality. Ultrasound of the abdomen was also normal. Total body fluorodeoxyglucose-Positron-Emission-Tomography (FDG-PET) scan was ordered and reported as FDG avid disease at the level V (posterior triangle) cervical lymph node on the right side, where the swelling was located (Figure 3). The area showed an SUV (standardized uptake value) of 8.83, favoring malignancy lesion.
The patient was treated as metastatic neck cancer with unknown primary. Right modified radical neck dissection was performed. Intraoperatively, there was a presence of single mass in the right posterior triangle of the neck which was attached to the underlying anterior border of the trapezius muscle. The right accessory nerve was removed as it lies within close proximity to the mass. The right internal jugular vein and right sternocleidomastoid, the right submandibular and sublingual glands, together with fat tissues in levels I-V were removed. Postoperative period was uneventful. Histopathological examination of the right level V swelling showed masses of ‘ghost’ cells with foci of calcification, clusters of basaloid cells and scattered multinucleated giant cells (Figure 2B). No lymph node was identified. The calcified nodule is consistent with pilomatrixoma.

Figure 1. Clinical photography demonstrating the appearance of a swelling in the posterior triangle of the neck on the right side

Figure 2. (A) The fine-needle aspiration smear with cytologic features mistaken for carcinoma, including sheets of cells with pleomorphic, hyperchromatic nuclei, prominent nucleolus and moderate cytoplasm. (Papanicolaou ×200) (B) Histopathological examination of the swelling showed masses of ghost cells with clusters and lobules of basaloid cells. (H&E ×200)
Figure 3. (A) CT scan showed a calcified nodule in the right posterior neck (arrow). (B) FDG-PET scan revealed a hypermetabolic lesion is observed in the mass.

**DISCUSSION**

In 1961, Forbis and Helwing introduced the term pilomatrixoma, a benign hair matrix tumour historically known as ‘calcifying epithelioma of Malherbe’. The incidence of pilomatrixoma is rare, accounting for 1.04% of all benign skin lesions. The tumour primarily affects children and adolescents, with a slight female preponderance. It usually manifests as a solitary, asymptomatic, slow growing, and hard subcutaneous nodule. Due to the benign nature of this disease, simple excision of the lesion is the treatment of choice. It is associated with a very low recurrence rate after surgery.

In all cases of neck swelling, the attending doctors need to ascertain whether this is essentially a malignant or a benign lesion. Among the clue from the examination to suggest predilections towards malignancy are the hard consistency of the swelling and fixation to the underlying skin. Our patient presented with these features. Together with the background family history of nasopharyngeal carcinoma, a malignant lesion needs to be ruled out. In the current otolaryngologic teaching, the commonest and least invasive investigations to perform in cases of neck swelling are fine needle aspiration cytology and radiological imaging. Aspiration cytology is a useful diagnostic tool in cases of neck swelling, with good overall sensitivity and specificity. Incision biopsy provides more tissue; hence, gives a superior diagnostic accuracy compared to cytology. However, in cases of metastatic neck carcinoma, incision biopsy can breach the tumour capsule and cause tumour spillage to the skin and worsened the prognosis of the disease. Therefore, in our patient, cytology aspirates and a CT scan was the first line of investigations ordered.

The role of cytology in the diagnosis of pilomatrixoma is debatable. It is generally agreed that cytology can provide conclusive diagnosis of pilomatrixoma, when specific features are looked for. The presence of ghost (shadow) cells, basaloid cell clusters, multinucleated giant cells, and calcium deposits in the appropriate clinical setting permits diagnosis by aspiration. However, the distinction from other primary malignancies is extremely difficult, and reports show that pilomatrixoma has been very often misdiagnosed as carcinoma. A large, retrospective review on cases of pilomatrixoma showed that correct preoperative cytological diagnosis is very low even when it was done by the experienced cytopathologist. Among the common reasons for the diagnostic error were predominance of one cellular component over the others and non-representative aspirated material. In our case, the diagnosis from FNAC was metastatic neck malignancy. This was made based on the visualized sheets of cells with pleomorphic, hyperchromatic nuclei, prominent nucleolus and moderate cytoplasm, which was thought to represent malignant cells. Therefore, the diagnosis of pilomatrixoma is best established from incisional biopsy or histopathology specimens, rather than FNAC.

In our case, following the cytological diagnosis of metastatic carcinoma of the neck node, the patient underwent conventional work-up to determine the primary origin. These investigations failed to show the ‘primary’ lesion. We then proceeded with a total-body FDG-PET scan, which showed positive results for malignancy. F-18 fluoro-2-deoxyglucose (FDG) is the most commonly used isotope for PET/CT. It is a glucose analogue that shows pathologic uptake in
tumour tissues due to over expression of glucose transport proteins. In one review article, the overall sensitivity, specificity, and accuracy rates of FDG-PET in detecting unknown primary tumors were 88.3%, 74.9%, and 78.8%, respectively. FDG-PET also detected 28.54% of tumours that were not detected by conventional work-up. Our case emphasizes that in cases of pilomatrixoma, FDG-PET scan gives a false-positive result, similar to one previous report in the literature. As a learning curve from our case, here are the steps that should have been taken to reduce the diagnostic error in cases of hard mass in the neck. Firstly, the doctor should know that pilomatrixoma is one of the differential diagnoses of a hard subcutaneous mass in the neck. When asking for a FNAC, always communicate with the cytopathologist of this suspicious diagnosis and let them aware and look vigilantly for the features of pilomatrixoma in the specimens. Performing an incisional biopsy to investigate a hard mass in the neck can give superior diagnostic value compared to FNAC, but be aware that once the diagnosis of carcinoma, proper surgery is needed quite urgently to reduce tumour spill and worsen the prognosis. Another step, when available is to perform a frozen section biopsy when doing the surgery, whenever the diagnosis is vague. This can minimize the unnecessary extensive surgery. Intraoperatively, if the diagnosis is still in doubt, a more conservative and minimal resection is an appropriate option and vital structure like accessory nerve should be preserved if it is distant from the primary lesion.

CONCLUSION

Pilomatrixoma is a benign tumour that may be misdiagnosed as a carcinoma, resulting in unnecessary aggressive therapy. Cytopathologist needs to be reminded of the potential diagnosis for them to look for specific features when examining the specimens. PET scan can give a false-positive result for malignancy in case of pilomatrixoma. Pilomatrixomas must be included in the differential diagnoses of hard subcutaneous lumps in the neck.

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REFERENCES