CASE REPORTS OF A RARE NEOPLASM

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INTRODUCTION

This article emphasises the need to approach the routine 'lumps and bumps' with caution. Two cases presented here were referred to the surgeons by general practitioners. The lumps had the appearance of common tumours but later turned out to be rare and aggressive neoplasms. This re-establishes the age-old dictum of medicine that nothing is routine in surgical practice.

This article describes two different presentations of an extremely rare cancer.

Case 1

A 97-year-old lady, self-caring, fit and healthy with no significant past medical history, was referred to the surgical clinic with a slow-growing fungating lump above the right knee of six months' duration. On examination it was 6 x 4 cm, non-tender, fixed to the skin, free from underlying structures and with an offensive odour. No nodal involvement in the inguinal region was detected.

Knee radiographs showed intact bone. CT scan revealed a mass separate from the bone, possibly arising from the skin. The blood profile was normal. The radiologist's impression was chondro- or fibro-sarcoma and the patient was referred for an orthopaedic review.

The patient was admitted to orthopaedics and a biopsy was taken. Histology revealed an infiltrating merkel cell carcinoma (MCC) (see figures) and the patient was referred to the regional plastic centre where surgical removal of the tumour and skin grafting were undertaken. The wound healed satisfactorily.

No further steps were taken considering the age and lifestyle of the patient and to date she is still alive six months after the operation.

Case 2

A 51-year-old gentleman with multiple medical problems including extreme obesity, hypertension and recurrent renal stones was referred by his GP to the surgical clinic with a nodule in the left little finger and a presumptive diagnosis of pyogenic granuloma. The lump rapidly increased in size and bled profusely. It had to be removed surgically. Biopsy showed merkel cell carcinoma and resection margins were free from the tumour indicating a complete resection.

The patient was kept under periodic review until about three years later when he showed ipsilateral axillary lymph node enlargement which showed metastatic merkel cell tumour on excision biopsy. He was promptly referred to the regional oncology centre for radiotherapy. In the following year he had two pathological fractures of the humerus and a

bone scan suggested metastasis to the bone. The fractures were internally fixed and he was considered for chemotherapy to arrest the progression of the disease.

At present, four years after chemotherapy, he is still in remission.

Figure 1 Merkel cell carcinoma (low power)
Image courtesy of Path Labs, Furness General Hospital

DISCUSSION

Merkel cells are mechanoreceptors⁽¹⁾ belonging to the amine precursor uptake and decarboxylation cell (APUD) family⁽²⁾ and are situated in the basal layer of the epidermis. These give rise to a rare neuroendocrine type of tumour related to small cell lung carcinoma, carcinoid tumours and the tumours derived from the neural crest⁽³⁾.

The aetiology is unknown but sun exposure^(1,4), chromosomal abnormalities⁽⁴⁾ and immunologic mechanisms are thought to be the causative factors. MCC has been found to occur in immunosuppressed patients and cures have been described with the use of tumour necrosis factor (TNF) alpha and interferon⁽⁴⁾.

Figure 2 Merkel cell carcinoma (high power)
Image courtesy of Path Labs, Furness General Hospital

The incidence of the disease is not known, due to its extreme rarity⁽⁴⁾ and about 875 cases⁽¹⁾ have been described so far in the literature between Toker's first report in 1972 and November 2000. The male:female ratio is 1.5:1⁽¹⁾. This is primarily a disease of the aged population (median 68 years, range 15-97 years)⁽⁴⁾.

Clinically it is a rapidly-growing, painless, non-tender, shiny, bluish red, intracutaneous nodule^(1,4) in which the clinical diagnosis can be confirmed by histology and immunohistochemical methods. These show positivity for low molecular weight keratin, neurofilaments and neuron-specific enolase⁽¹⁾. This feature helps to diagnose undifferentiated dermal tumours and small cell lung carcinoma⁽⁴⁾. There are several other minor markers. Under electron microscopy, cytoplasmic peripheral dense core granules and para-nuclear fibrous bodies help to distinguish MCC from oat cell carcinoma⁽¹⁾.

Staging is done following the model established by Yiengpruksawan *et al*⁽⁵⁾. Stage I is local disease (1a <2 cm and 1b >2 cm). Stage II is lymph node positive and Stage III is distant metastasis⁽¹⁾. At presentation up to 89% are at stage I⁽⁴⁾. The tumour commonly affects the head and neck region (47%) and extremities (47%)⁽¹⁾.

Confusion exists in establishing the best treatment guidelines. Surgically, wide excision with 3cm or greater margins is advocated, except in the head and neck region where Mohs micrographic surgery is advocated for cosmesis^(1,6). Radiotherapy has been used both primarily and as post-surgical irradiation to the local area and regional lymph node region to achieve better control of recurrence^(1,7). Chemotherapy can also be used as adjuvant therapy, treatment of loco-regional diseases and distant metastasis⁽⁴⁾.

At present the five-year survival rate is as low as 21-30%⁽⁷⁾. Prognostic chromosomal markers are being researched, which if identified will guide the management in future.

CONCLUSION

- The diagnosis of 'lumps and bumps' should be approached with an open mind because they are common presentations for both common and rare tumours.
- Case 2 highlights the importance of sending all excised materials for histology.
- Patients with rare cancers should be referred for specialist review to regional oncology institutes immediately following diagnosis.

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