CASE REPORT

An unusual breast mass: primary synovial sarcoma

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vicky.doyle@doctors.org.uk**SUMMARY**

We describe a 54-year-old woman presenting with mastalgia and a 6 mm breast lesion on imaging. Core biopsy revealed a lesion characterised by a predominant epithelioid and a minor spindle cell component. Our differential diagnosis included intraduct papilloma/adenoma and adenomyoepithelioma. However, initial immunohistochemistry did not support these diagnoses and further immunohistochemistry raised the possibility of a synovial sarcoma. This was confirmed with the finding, using fluorescence in-situ hybridisation, of the characteristic translocation $t(x; 18) (p11.2; q11.2)$. Establishing a diagnosis of synovial sarcoma at unusual sites may be difficult, especially when limited tissue is available, for example, within a core biopsy. In this case, immunohistochemistry was useful, but cytogenetics was the key additional investigation. It is important to consider the possibility of rare tumours when the morphological and immunohistochemical features of a lesion initially appear conflicting or inconclusive.

BACKGROUND

Synovial sarcomas can arise in almost any soft tissue location although it is rare for them to invade the actual joint space. Synovial sarcomas tend to arise in young adults, typically 15–40 years of age.¹ They are often asymptomatic; however, pain is not an infrequent presentation as in our case.² Grossly, synovial sarcomas appear as a well-circumscribed, firm nodule, which is often greyish-pink in colour. Focal calcifications can be seen which can make them detectable radiologically.^{3–4} Synovial sarcomas have two distinct histological subtypes: the classic biphasic pattern and the less common monophasic pattern. Synovial sarcomas have a tumour-specific chromosome translocation $t(x; 18) (p11.2; q11.2)$ this leads to fusion genes *SYT-SSX1* and *SYT-SSX2*.⁴ Breast carcinoma is by far the most frequently reported tumour of the breast. We report a very unusual case of a primary synovial sarcoma of the breast in a 54-year-old woman in which the core biopsy was initially suggestive of a more common breast lesion such as an intraduct adenoma/papilloma or adenomyoepithelioma.

CASE PRESENTATION

A 54-year-old woman presented with left-sided mastalgia. Mammography revealed a possible cyst in the left lower outer quadrant and an ultrasound scan revealed a 6 mm complex cyst. Fine needle aspiration cytology was inconclusive, showing only atypical epithelial cells. Core biopsy was therefore obtained.

Microscopic examination of the core biopsy showed part of a lesion characterised by a florid epithelioid and a minor spindle cell component (figure 1A). There was evidence of mild to moderate

cytological atypia and conspicuous mitoses. The initial differential diagnosis included an intraduct papillomatous lesion, a duct adenoma with epithelial atypia and an adenomyoepitheliomatous lesion. Immunohistochemistry demonstrated that the epithelioid component expressed epithelial membrane antigen (EMA) (figure 1B) and cytokeratin (CK8) (figure 1C). The spindle cell component was not myoepithelial in nature—negative for α -smooth muscle actin (figure 1D) and smooth muscle myosin heavy chain. The spindle cell component was also weakly EMA positive. Oestrogen receptor and p63 showed no expression within either the spindle or epithelioid components. The initial panel of immunohistochemical stains did not support our initial morphological differential diagnoses and raised the possibility of the lesion being a carcinosarcoma, a sarcomatoid carcinoma or possibly a synovial sarcoma.

Additional immunohistochemistry for CD99 and Bcl-2 (figure 1E,F) was non-contributory in either confirming or refuting the diagnosis of synovial sarcoma. However, fluorescence in-situ hybridisation analysis revealed a *SYT (SS18)* rearrangement indicating the presence of a $t(x; 18)$ translocation and characteristic of a synovial sarcoma.

TREATMENT

The patient was treated with a wide local excision, and pathological examination of the mass demonstrated a well-circumscribed 8 mm synovial sarcoma with clear margins. Further investigations demonstrated neither evidence of metastatic spread nor identification of an alternative primary site.

OUTCOME AND FOLLOW-UP

One year on, the patient remains clinically well with a surveillance chest radiograph showing no evidence of disease recurrence.

DISCUSSION

Primary synovial sarcoma of the breast is a rare tumour with relatively few reported cases. Synovial sarcoma can arise in almost any soft tissue location although it is rare for it to invade the actual joint space (as is implied by the name). Synovial sarcoma tends to arise in young adults, typically 15–40 years of age.¹ They are often asymptomatic; however, pain is not an infrequent presentation as in our case.² Grossly, synovial sarcomas appear as a well-circumscribed, firm nodule, which is often greyish-pink in colour. Focal calcifications can be seen which can make them radiologically detectable.^{3–4}

Synovial sarcoma shows two main histological subtypes. There is the classic biphasic pattern and

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Figure 1 Histological appearances of the tumour: (A) H&E, (B) epithelial membrane antigen, (C) cytokeratin 8, (D) α -smooth muscle actin, (E) CD99 and (F) Bcl-2.

the less common monophasic pattern. Both patterns contain spindle cells arranged in fascicles, with tapered nuclei and pale cytoplasm within a collagenous background stroma. Mitoses may be quite frequent. In addition, in the biphasic pattern, the epithelial component contains glandular structures lined by either cuboidal or columnar epithelium. A poorly differentiated subtype can also be seen.⁴

Synovial sarcomas show at least focal expression for epithelial markers such as EMA and cytokeratin. One-third of cases are S100 positive. Two-thirds of cases are CD99 and Bcl-2 positive. They are negative for CD34.⁴ Lack of expression of Wilms tumour protein-1 can be useful in distinguishing synovial sarcoma from an epithelial mesothelioma.⁵

The initial core biopsy in our case showed part of a lesion, which was characterised by a florid epithelioid and a minor spindle cell component. More common breast lesions with a biphasic profile include duct papilloma, intraduct papilloma or an adenomyoepithelioma. The site of tumour (breast) and limited tissue (core biopsy) in this case made consideration of a rare synovial sarcoma more challenging. Immunohistochemistry was useful in demonstrating that the tumour showed no myoepithelial differentiation and prompted consideration of a sarcoma.

Synovial sarcoma has a tumour-specific chromosome translocation $t(x; 18) (p11.2;q11.2)$ which leads to fusion genes *SYT-SSX1* and *SYT-SSX2*. The *SYT-SSX1* fusion gene is often seen in conjunction with the biphasic pattern whereas the *SYT-SSX2* fusion gene is often associated with the monophasic pattern.⁴ The identification of the tumour-specific translocation has enabled molecular testing as a means of diagnosis. This is particularly useful when morphology and immunohistochemistry are suggestive of the diagnosis, but not conclusive.¹

Synovial sarcoma is an aggressive tumour with a high incidence of metastases, particularly to lung and lymph nodes. Different series show wide ranges in 5-year survival rates (24–76%).^{6–7} One study examining prognostic factors on 128 patients showed that as well as the monophasic subtype, that tumour necrosis and mitotic index were also important high-risk indicators and that these factors should be used to guide more aggressive management.

This case of primary synovial sarcoma of the breast illustrates a rare cause for a breast mass, in which the diagnosis was not suspected until the examination of core biopsy. As there are many commoner lesions of the breast, which have an epithelial and spindle cell component, making a diagnosis of synovial sarcoma at this site may be difficult. In our case, immunohistochemistry was useful in excluding the presence of myoepithelial

cells, but it was of limited value in classifying the sarcoma. Cytogenetics is therefore an important adjunct to diagnosis. This case demonstrates the importance of considering rare tumours when the morphological and immunohistochemical appearances are initially inconclusive—and the utility of techniques such as cytogenetic analysis as a means of reaching a precise diagnosis.

Learning points

- ▶ Primary synovial sarcoma of the breast is rare, with relatively few reported cases.
- ▶ It is important to consider lesions that may be rare at the site in question when the morphological features and immunohistochemical profile of a lesion initially appear inconclusive.
- ▶ Cytogenetics is an important adjunct, and where positive, very often enables a precise diagnosis.

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Competing interests None.

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