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CASE REPORT

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Clonal chromosomal changes in juxta-articular myxoma

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Abstract Cytogenetic analysis of a juxta-articular myxoma revealed two distinct cytogenetically abnormal cell populations: inv(2)(p15q36) and +7, t(8;22)(q11–12; q12–13). These clonal chromosomal changes, the first to be reported in this tumour type, suggest that at least some juxta-articular myxomas are neoplastic rather than reactive in nature.

Key words Myxoma · Juxta-articular · Cytogenetics

Introduction

Myxomas are localized, paucicellular growths of fibroblast-like cells in an abundant myxoid matrix [6]. The myxoid aspect of the matrix is related to the presence of excessive amounts of glycosaminoglycans. According to the clinical setting, different types of myxoma are defined: cardiac, intramuscular, cutaneous, juxta-articular myxoma and myxoma of the jaw [3]. Juxta-articular myxomas usually occur in adult men, and the knee joint is most frequently involved. In contrast to cardiac and cutaneous myxomas, juxta-articular myxomas are not associated with other syndromes or tumours; they are often associated with trauma and are considered to be an exuberant reactive fibroblastic proliferation with overproduction of mucin [3]. In fact, according to the WHO definition, all soft tissue myxomas are regarded as 'probably nonneoplastic' [10]. Nevertheless, in their original paper on juxta-articular myxoma, Meis and Enzinger admit that some cases may be neoplastic owing to the large size and the diffuse infiltrative pattern in some lesions [5].

This is the first report on a juxta-articular myxoma with clonal chromosomal changes; it suggests that at least some of these lesions are neoplastic in nature.

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Clinical history

A 51-year-old woman complained for 1 month of some pain at the medial and dorsal side of the left proximal forearm and elbow, which was consistent with a "golfers' elbow". She was treated with oral nonsteroidal anti-inflammatory drugs, which relieved the pain to some extent. About 3 weeks later she suddenly felt a swelling at the volar side of the proximal forearm A mobile palpable mass of 3 cm was noted. The tumour was lobulated and hypo-/hyperintense on T1/T2-weighed MRI imaging, and was localized on the anterior side of the supinator muscle and radius immediately lateral to the insertion of the biceps tendon. After gadolinium administration there was only some signal increase at the periphery and some septum-like staining at the deep side of the mass. Peroperatively, the tumour was well demarcated, but on the deep medial side there was some extension proximally along the biceps tendon. The wound healed uneventfully, and 9 months after resection no recurrence was documented.

Materials and methods

The resection specimen was received fresh. A tissue fragment was processed for culturing and cytogenetic analysis according to procedures previously described [4]. The remaining tissue was fixed in formalin (6%) and processed to paraffin.

Results

Macroscopic analysis revealed a soft, well-delineated whitish nodule. On histology, the lesion was multinodular and myxoid, with spindle-shaped to stellate fibroblast-like cells embedded in the myxoid matrix. Tumour cell nuclei were regular and euchromatic. In between the

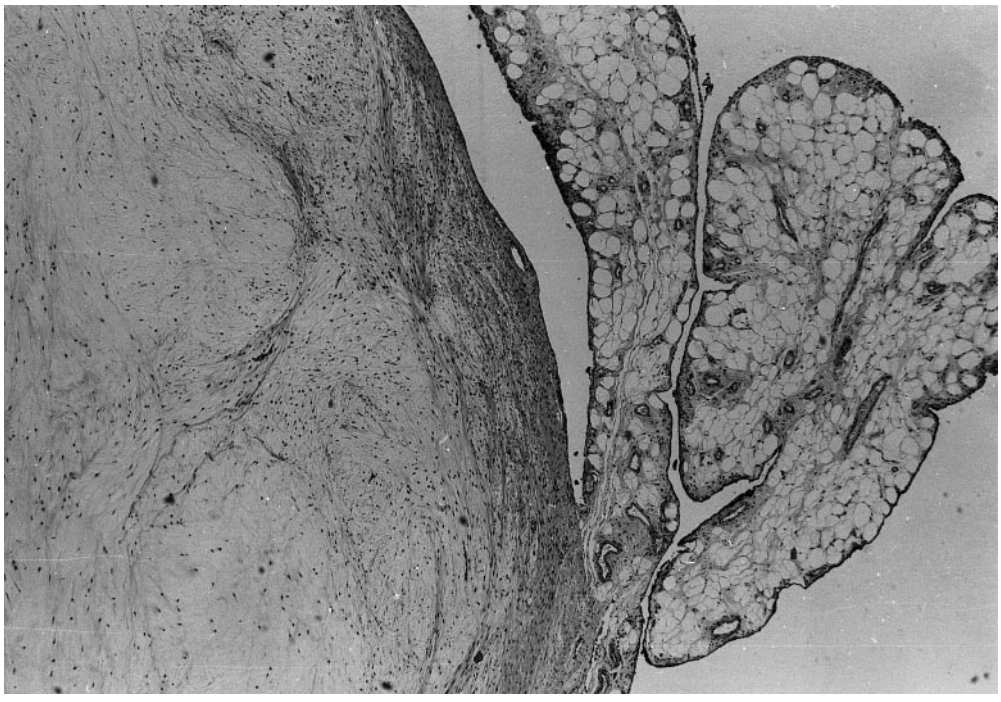


Fig. 1 Juxta-articular myxoma. Note the vague lobularity of this myxoid tumor and the attachment to synovial villi. Haematoxylin & eosin, ×125

nodules there was more cellular and collagenous tissue. The mass was poorly vascularized throughout. Focally, synovial tissue was attached to the tumour (Fig. 1). There was no associated cyst.

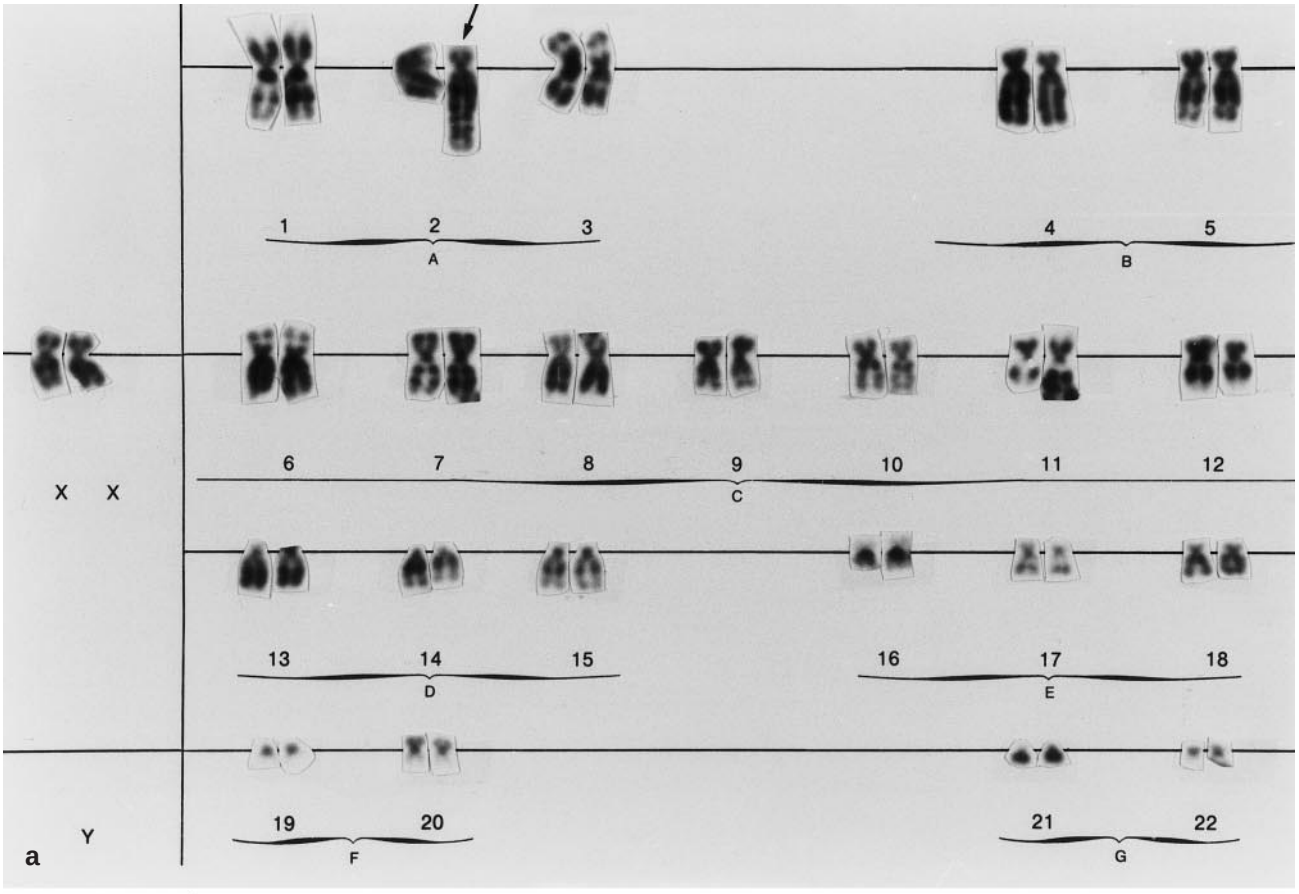
Cytogenetic analysis was performed on 20 G-banded metaphases obtained after 8-day culture. An inv(2)(p15q36)was found in 2 cells and a 47, XX, +7, t(8;22)(q11–12;q12–13) karyotype was found in 3 other metaphases (Fig. 2a, b). A normal female karyotype was present in the remaining 15 metaphases.

Discussion

In soft tissue tumours, myxoid changes are not unusual. This appearance can be a secondary change, as in neurofibroma, lipoma, leiomyoma or dermatofibrosarcoma protuberans. However, it can be a hallmark of the tumour, as in myxofibrosarcoma, fibromyxoid sarcoma, myxoid liposarcoma, and extraskeletal myxoid chondrosarcoma [3]. The last two tumours show consistent chromosomal changes, t(12;16) or t(12;22) in myxoid liposarcoma and t(9;22) in extraskeletal myxoid chondrosarcoma [1, 8, 9]. A specific t(17;22) is found in dermatofibrosarcoma protuberans [7]. Among the benign myxomas, only cardiac myxomas have been reported to show clonal chromosomal changes. Cytogenetic investigations of ten cardiac myxomas revealed that telomeric associations were frequently observed, together with involvement of the short arm of chromosome 12 in clonal and nonclonal abnormalities (for review, see Dijkhuizen et al. [2]).

This is the first report on juxta-articular myxoma with clonal chromosomal changes, and this juxta-articular myxoma is also the first we have been able to karyotype. The clonal chromosomal changes, which will hopefully be confirmed in future cases, suggest that at least some of these lesions could be neoplastic rather than reactive in nature. The fact that juxta-articular myxomas are apt to recur when incompletely resected might support this hypothesis. Nevertheless, it is becoming clear that the presence of clonal chromosomal changes does not always mean neoplasia per se. The finding of clonal chromosomal changes in various kinds of tissues and lesions that are not unequivocally regarded as neoplastic challenges this old dogma.

Fig. 2 G-banded karyotypes showing **a** an inverted chromosome 2 (*arrow*) and **b** an extra chromosome 7 plus a translocation between chromosomes 8 and 22 (*arrows*)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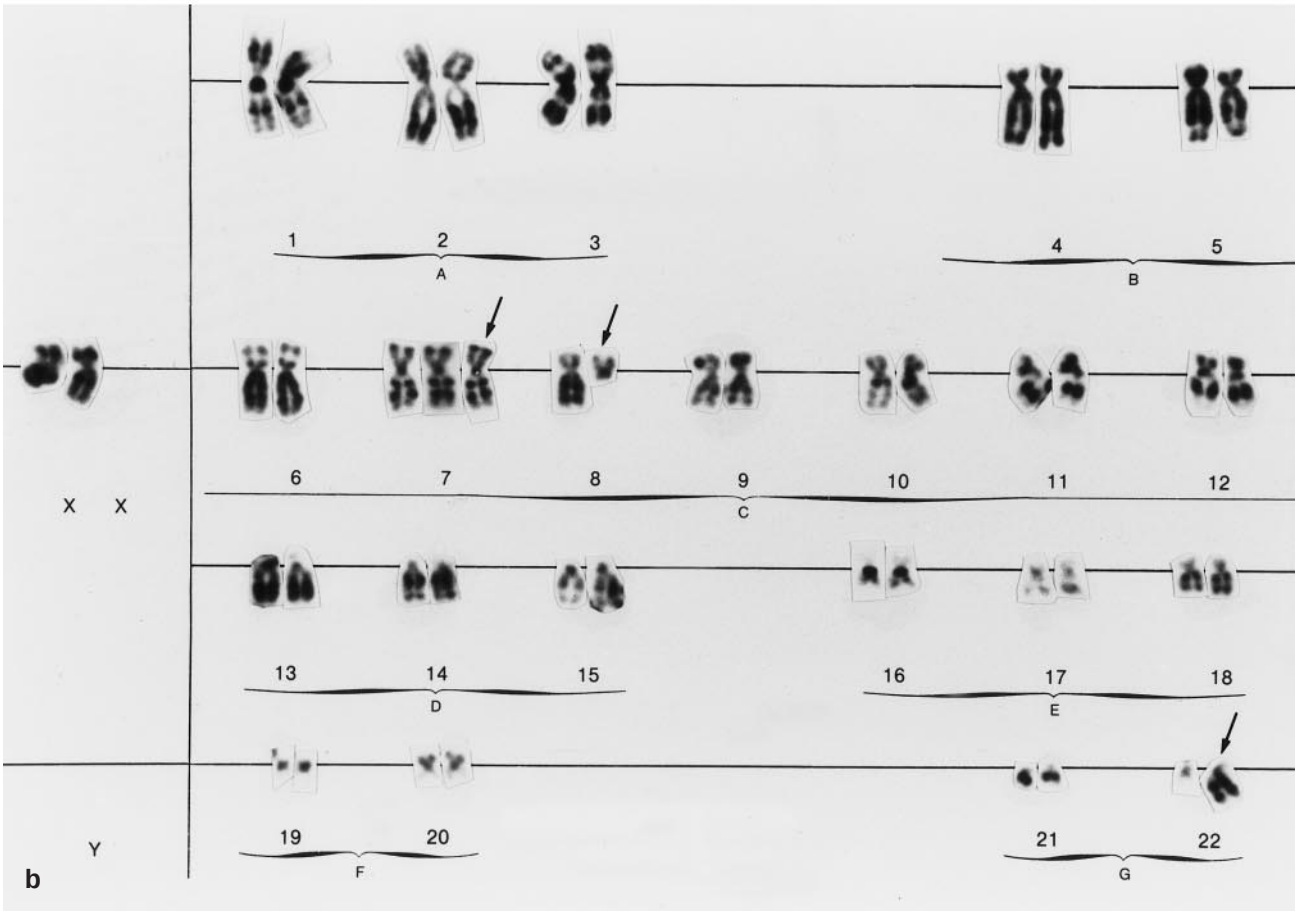
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