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CASE REPORT

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Synovial chondromatosis: clonal chromosome changes provide further evidence for a neoplastic disorder

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Abstract Synovial chondromatosis is a rare lesion, which is still believed by most authors to be reactive rather than neoplastic. We report on a case of synovial chondromatosis with clonal chromosomal changes [43,XX,der (1) t (1;13) (p21–22;q21),-6,-13,-14, add(21) (q21)]. The presence of clonal chromosomal changes in this and in three previously reported cases suggests that synovial chondromatosis is a true neoplastic lesion.

Key words Synovial chondromatosis · Chromosomes · Karyotype

Introduction

Synovial chondromatosis is a rare disorder characterized by the formation of multiple benign cartilaginous nodules in the synovium. The knee joint is the predilection site, followed by the hip and elbow. The disease affects males twice as often as females, with a peak incidence in the fifth decade [3].

Synovial chondromatosis is considered by most authors to be a reactive, non-neoplastic process. Nevertheless, these lesions can be quite cellular and can display nuclear atypia. In addition, there is a tendency for recurrence and there are reports of patients with synovial chondrosarcoma who had synovial chondromatosis many years before [3]. We discuss a case of recurrent synovial chondromatosis with clonal chromosomal changes. This finding lends further credence to the idea that synovial chrondromatosis is a true neoplastic lesion.

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Clinical history

A 57-year-old female patient complained of pain and loss of flexion in the left knee. MRI was consistent with synovial chondromatosis. At arthroscopy, a synovial nodule was removed and the hypertrophic synovium was shaved. After a temporary improvement, a recurrence was documented. An open synovectomy was performed with removal of the abnormal tissue. By 5 months after surgery, the patient had regained full extension with no evidence of recurrence.

Materials and methods

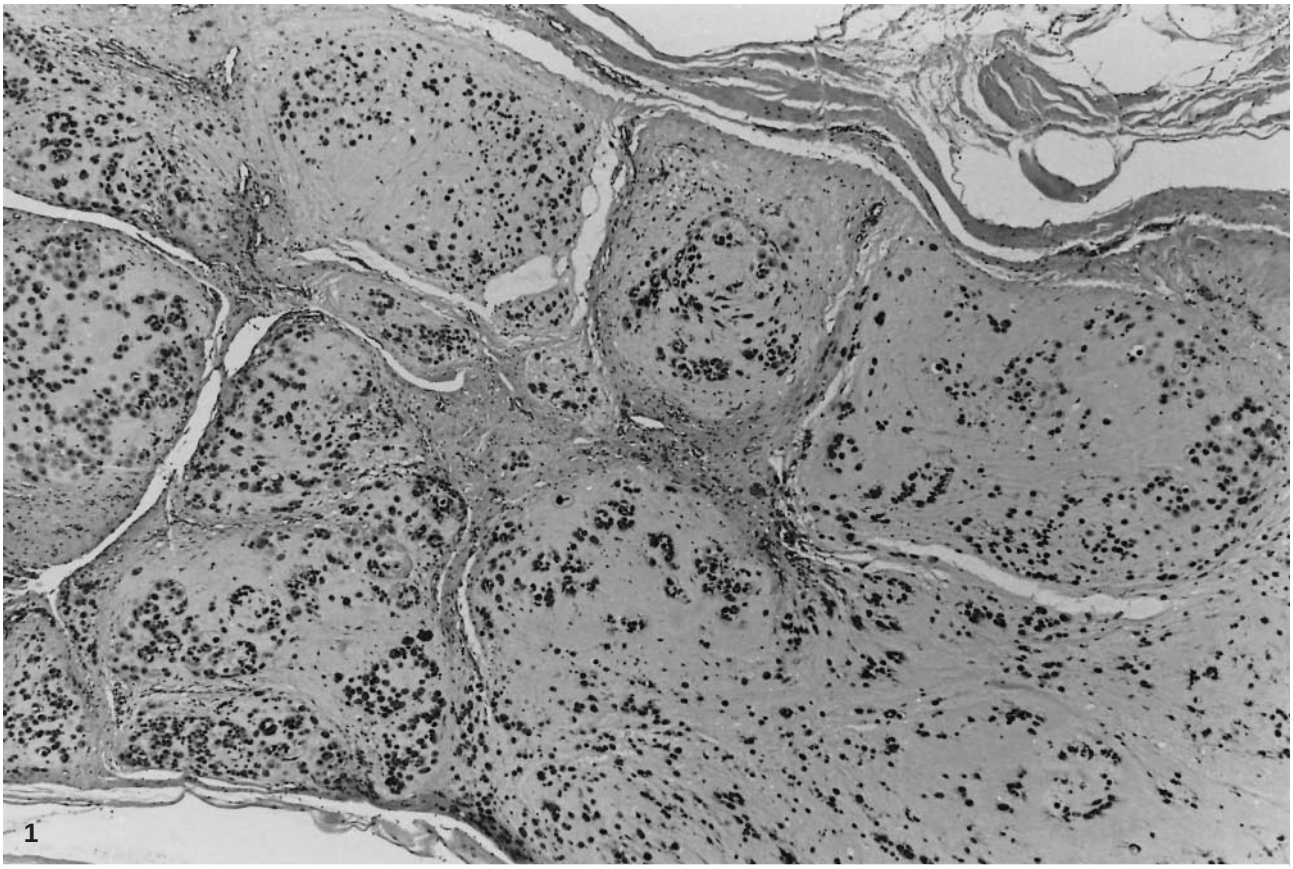
The major part of the resection specimen was fixed in formalin (6%) and processed to paraffin. A sample of the tissue was disaggregated overnight by collagenase treatment and cultured for 4 days for cytogenetic investigation according to the standard procedure used in our laboratory [5]. The karyotypic descriptions and the clonality criteria follow the recommendations of ISCN [7].

Pathologic findings

On histology, the lesion was found to have a typical nodular architecture. The size of the lobules varied considerably. Each nodule was composed of hyaline cartilage with increased cellularity at the edge of the nodule (Fig. 1). Chondrocytes with an enlarged pleomorphic nucleus were present in the areas of increased cellularity. There was no necrosis or myxoid change in the stroma. The primary and recurrent lesions showed an identical morphology.

Most (13) of the 20 G-banded metaphase cells analysed exhibited the following karyotype:

43, XX, der (1) t (1;13)(p21–22;q21), -6,-13,-14, add(21) (q21) (Fig. 2).



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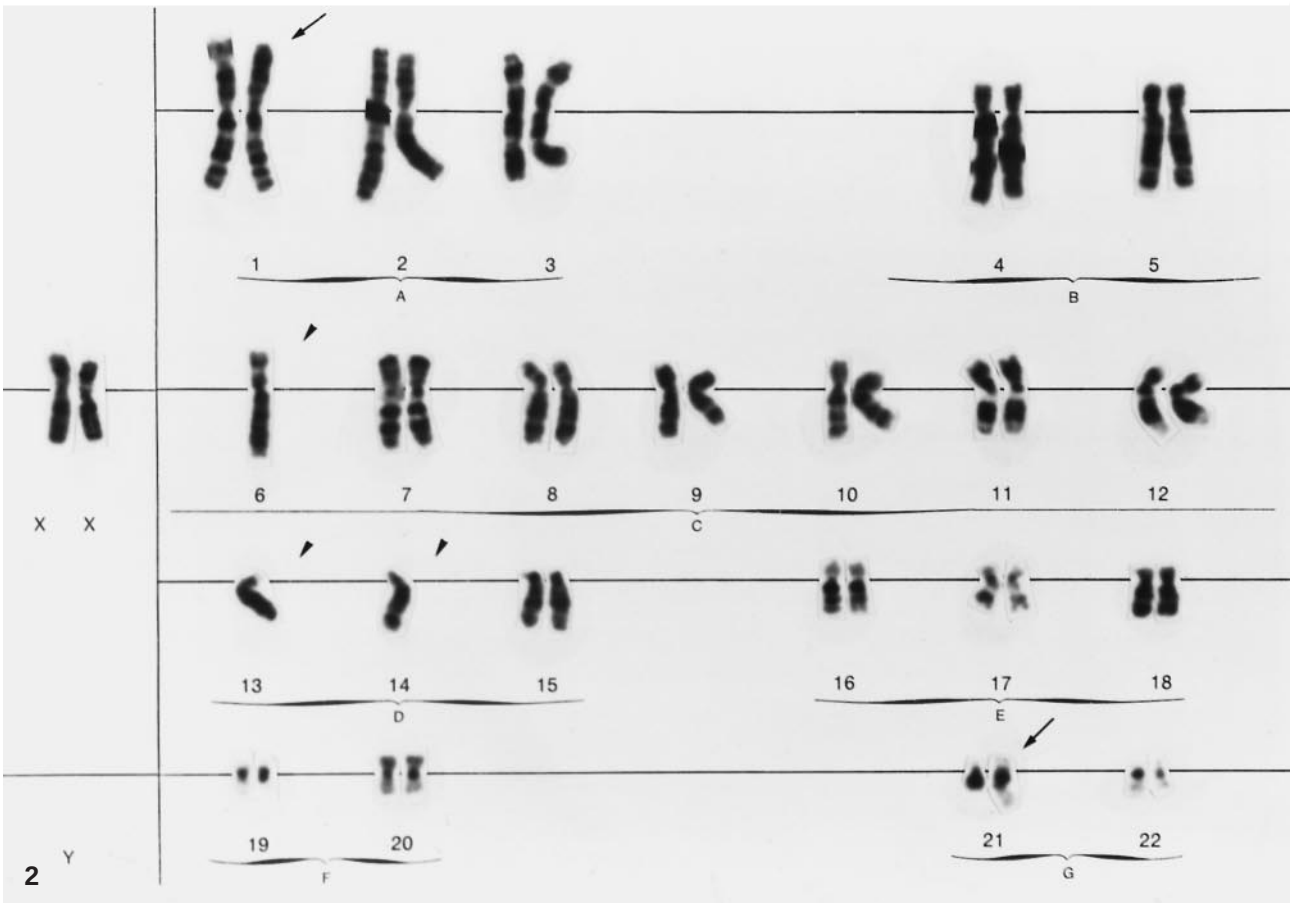

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Table 1 Clinical and cytogenetic findings in four synovial chondromatous lesions investigated (cases 1–3, see [6]; case 4 see present report)

Case no./ age/sex	Joint	Karyotype
1/46/M	Knee	44,XY,add(1)(p36),del(1)(p13p22),add(6)(p25),del(7)(q22q32), del(10)(q21),add(11)(q13),-17,-18[9]/46,XY[36]
2/47/M	Hip	47,XY,der(1)inv(1)(p13q25)del(1)(q25q32),t(1;12)(q25;q13),+5, er(12)add(12)(p11)t(1;12)(p22;q13)[8]/46,XY[15]
3/72/M	Temporo-mandibular	47,XY,add(10)(q26),+20[12]/46,idem,-6[5]/46,XY,t(2;4)(q33;q21), add(21)(p11)[4]
4/58/F	Knee	43,XX,der(1)t(1;13)(p21–22;q21),-6,-13,-14,add(21)(q21)[13]/46,XX[7]

Discussion

Cytogenetic information is available on six synovial chondromatous lesions: five synovial chondromatoses and one synovial chondroma. Four of them (3 synovial chondromatoses, 1 synovial chondroma) exhibited clonal chromosome abnormalities [6]. The case of synovial chondromatosis reported here does not share any chromosome aberrations with the three cases previously reported (Table 1).

With regard to other chondromas, 8 of the 22 (enchondroma, periosteal chondroma and soft tissue) cases reported showed chromosome abnormalities (for review see [2]).

Structural abnormalities of chromosome 6 and rearrangements of band 12q13 seem to be the most frequent chromosome changes involved in chondromatous lesions [8]. Interestingly, the gene for the 2 chain of collagen type XI has been mapped to the short arm of chromosome 6, band p21.3 [4] and the gene for type II collagen, the major constituent of normal cartilage, to chromosome 12 at 12q13 [10]. Whether these genes are involved in tumorigenesis is not known.

Synovial chondromatosis should be differentiated from the much rarer synovial chondrosarcoma. According to Bertoni et al., synovial chondrosarcoma shows a sheet-like growth pattern with rounding of cells, necrosis and myxoid change in the stroma [1]. Cytogenetic analysis is not very helpful in this regard since, with the exception of extraskeletal myxoid chondrosarcoma, which is characterized by a t(9;22)(q34;q12) [9], a specific chromosomal abnormality has not been yet identified in classical chondrosarcomas [8].

◀ **Fig. 1** Mature hyaline cartilage nodules are embedded in synovial tissue. H&E, ×250

Fig. 2 G-banded karyotype showing der(1)t(1;13)(p21;q21), add(21)(q21)(*arrow*) and loss of chromosomes 6, 13 and 14 (*arrowheads*)

In conclusion, the presence of clonal chromosomal changes in the four published synovial chondromatosis cases suggests that this lesion is neoplastic rather than reactive in nature.

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