Desmoplastic fibroma of the rib with cystic change

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Abstract

Desmoplastic fibroma (DF) is a rare, locally aggressive bone tumour, first described by Jaffe in 1958. DF earned its appellation as a result of its morphological resemblance to desmoid-type fibromatosis. The majority of DFs occur in the long bones, mandible and ilium. Presentation in the rib is extremely rare with only six cases reported previously. We report a case of a 42-year-old woman with DF of the rib. Microscopically the tumour was paucicellular comprising bland slender stellate to spindle cells, embedded in abundant loose myxocollagenous stroma. In addition, focal cystic change was noted. There was no specific immunohistochemical staining and importantly B-catenin stain showed a predominantly cytoplasmic staining. DF is associated with an indolent clinical course if adequately excised. As these tumours share morphological similarities with other fibrous/fibro-osseous tumours of bone and soft tissue, awareness is crucial to avoid misdiagnoses.

Introduction

Desmoplastic fibroma (DF) is a rare benign, locally aggressive bone tumour, first described by Jaffe in 1958.[1] DF shares a morphological overlap with desmoid-type fibromatosis. As such, it is widely regarded as the osseous manifestation of the latter. The vast majority of DFs occur in the metaphyseal region long bones, mandible and ilium. A small number has been reported in other location, including the scapula,[2] cranio-facial location,[3,4] toes[5] and spine.[6] However, presentation in the rib is extremely rare with only six cases reported in the past. Herein, we present the radiologic and pathologic features of a case of DF arising from the 7th rib, and briefly outline the treatment based prognostic implications of these findings.

Case report

A 42-year-old woman presented with a one-year history of a tender swelling over the left chest wall. Computed tomography revealed an intramedullary lytic lesion in the 7th rib with cystic change. The overlying cortex was intact. The lesion was completely resected. Macroscopic examination revealed a well circumscribed, 3.4 × 2.5 × 1.0 cm, fibro-gelatinous lesion with cystic change in mid portion of the rib, confined to the medullary cavity. Histopathological examination revealed a paucicellular lesion comprising slender, stellate to spindle shaped cells, embedded in an abundant loose myxocollagenous stroma. In addition, focal cystic change was also noted. (Figure 1a–c). The overlying bone is markedly thickened with remodelling, prominent osteoblastic rimming and woven bone apposition. Necrosis was not a feature and mitoses were absent. The lesional cells showed focal positivity for B-catenin, which was predominantly cytoplasmic (Figure 1d) and were negative for SMA and desmin. Based on the macroscopic, microscopic and immunohistochemical features a diagnosis of DF of bone was made.

Discussion

DF is a rare bone tumour, seen most frequently in adolescents and young adults.[7,8] The clinical presentation is highly variable and dependent on the site of presentation. However, pain is the commonest, initial, presenting symptom, as observed in our case. The majority of DFs have benign radiological features: lytic, well-demarcated lesions with sclerotic margins.[7–9] Occasionally, some lesions, particularly when larger in size, may exhibit more aggressive features including cortical destruction and soft tissue extension.[9] In addition, cystic change or degeneration, a feature also observed in our case, is an extremely rare finding. Radiologically, this can be a deceptive feature, resulting in a broad differential to include aneurysmal bone cyst, giant cell tumour or fibrous dysplasia.[9]

Microscopically, the majority of DFs are characterized by abundant collagenous matrix and a patternless arrangement of cytologically bland spindle to stellate cells, closely resembling desmoid type fibromatosis of soft tissue.[7,8] Cystic change is infrequently associated with DF.[9,10] DF shares morphological similarities with a wide variety of tumours (Table 1), chiefly desmoid type fibromatosis, fibrous dysplasia, osteofibrous dysplasia, low-grade fibrosarcoma of bone and low-grade osteosarcoma/low-grade paroosteal osteosarcoma.[11]

Desmoid type fibromatosis is morphologically similar to DF, comprising a proliferation of spindle cells arranged in broad fascicles. DFs can be mistaken for desmoid type fibromatosis, particularly when there is extension into the adjacent soft tissue. However, unlike desmoid type fibromatosis, the vast majority of DFs are negative for B-catenin. Hauben et al.[12] studied the involvement of the B-catenin pathway in DF and concluded that the B-catenin pathway did not play an integral role in tumourigenesis in DF, thus arguing against these two entities being biologically related.

Fibrous dysplasia is usually characterized by irregular, curvilinear trabeculae of bone dissected by a bland hypocellular fibroblastic stroma. While these irregular islands of bone do not feature in DFs, if samples are taken at the periphery of the lesion incorporating reactive bone or residual bone trabeculae, a diagnosis of FD can be erroneously made. However, in the absence of the characteristic ground-glass radiological appearance and GNAS gene mutation of fibrous dysplasia, a diagnosis of DF would be favoured.

Osteofibrous dysplasia, also a fibro-osseous tumour of bone, is almost exclusively seen in the cortical or medullary compartments of the tibia. It is composed of irregular fragments of woven bone,

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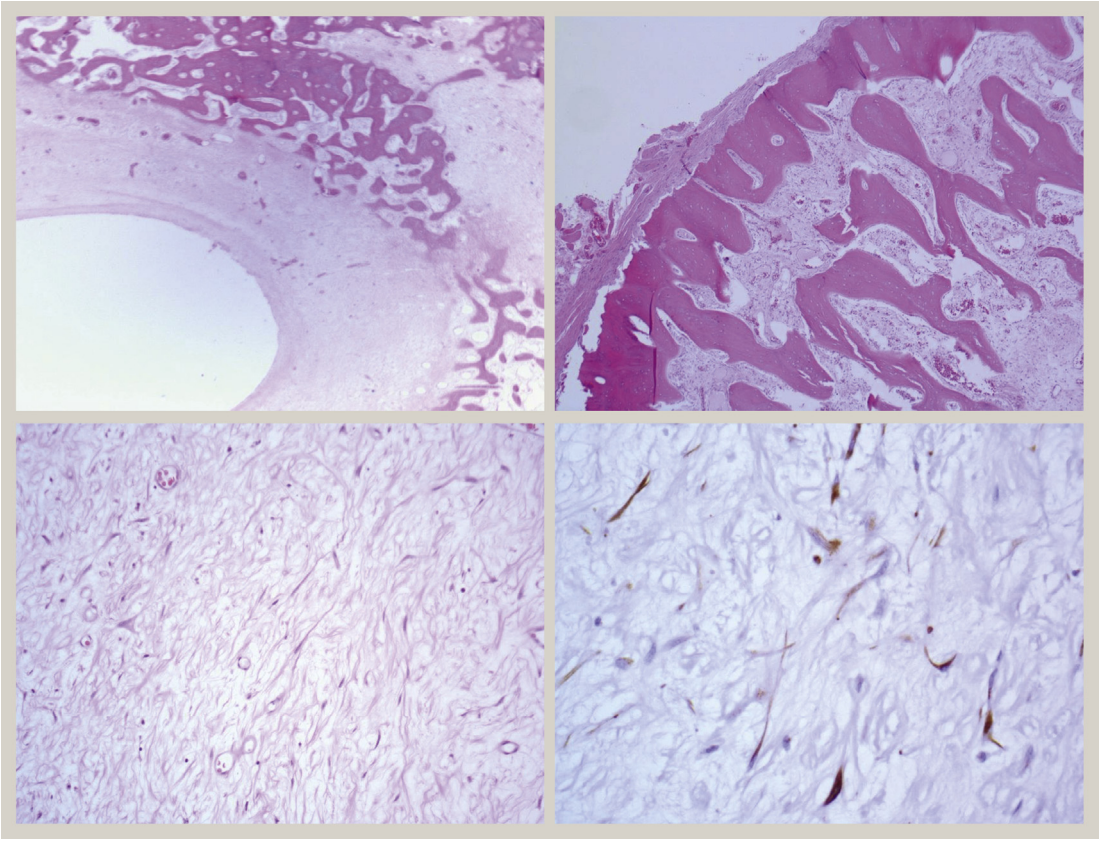

Figure 1 (**a**) scanning magnification of the tumour seen filling and expanding the rib with cystic change (H&E, ×16), (**b**) the overlying bone was thickened with evidence of remodelling (H&E, ×40) (**c**) the tumour was paucicellular comprising slender, stellate to spindle shaped cells, embedded in an abundant loose myxocollagenous stroma (H&E, ×200) (**d**) focal positivity for B-catenin with a predominantly cytoplasmic staining pattern (×400).

rimmed by osteoblasts and an intervening hypocellular spindle cell proliferation. In addition, scattered keratin-positive epithelial cells are also seen. In reality, the features of osteofibrous dysplasia are sufficiently distinct and should not be confused with DF.

Low grade fibrosarcomas are malignant neoplasms characterized by a uniform population of spindle shaped cells arranged in a fascicular or herringbone pattern with variable amounts of collagen production. These tumours have no specific immunophenotype and are usually a diagnosis of exclusion. Unlike DFs, they tend to show a greater degree of cellularity and mitotic activity, making the distinction readily apparent.

Low grade osteosarcoma is also characterized by irregularly shaped bony trabeculae with intervening fibroblastic proliferation, usually of varying cellularity, reminiscent of fibrous dysplasia; hence also a mimicker of DF, particularly if the latter exhibits overlying cortical destruction. Low-grade osteosarcomas, unlike DFs, tend to show a more permeative growth pattern, entrapment of host bone and the presence of *MDM2* amplification.[13] In addition, Crim et al.[11] reported three cases of DF occurring in the periosteum, which may radiologically and morphologically mimic parosteal osteosarcoma. Again, *MDM2* is of utility here as parosteal osteosarcomas also exhibit *MDM2* amplification.

It is also worth noting, that there have been three reported cases of DFs associated with extensive chondroid metaplasia, morphologically mimicking dedifferentiated chondrosarcoma.[14] In this scenario, the low cellularity of the spindle cell component and the absence of IDH1/IDH2 mutation would argue against a diagnosis of dedifferentiated chondrosarcoma.

DFs are considered neoplasms of intermediate biological potential, owing to their tendency to recur locally. A simple complete surgical excision is usually curative. However, reported recurrence rates range from 17 to 72%[8] which is due to the inclusion of all types of resection specimens from curettages to complete wide resection.[8] While a complete surgical excision is the ideal treatment option, the extent of the excision is often limited by the anatomical location of the tumour. This may be a prudent choice, particularly when radical procedures such as hemipelvectomies may result in a considerable reduction in the quality of life.[7]

In conclusion, DF is a rare, distinctive tumour of bone with an indolent clinical course following complete surgical resection. As these tumours share morphological similarities with other fibrous/fibro-osseous tumours of bone and soft tissue, awareness is crucial to avoid misdiagnoses. ◆