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Case Report

CASE REPORT

Fibrous dysplasia of rib presenting as a cystic mass in the lung

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Abstract

Fibrous dysplasia (FD) is a benign skeletal disorder that can affect one bone (monostotic form) or multiple bones (polyostotic form). It is a non-inherited bone disease, in which abnormal differentiation of osteoblasts leads to replacement of normal marrow and cancellous bone by immature bone with fibrous stroma. It is often asymptomatic and incidentally detected on radiographs. We report this rare disorder in a 22-year-old lady who presented to us with cough and breathlessness. Her chest radiograph showed a cystic mass extending into the chest wall. On computed tomography scan, mass turned out to be FD of the second rib. Histopathology of the lesion confirmed the diagnosis.

INTRODUCTION

Fibrous dysplasia (FD) is a non-inherited, skeletal developmental abnormality, in which abnormal differentiation of osteoblasts leads to replacement of normal marrow and cancellous bone by immature woven bone with fibrous stroma [1]. It can be monostotic, involving a single bone, or polyostotic, involving two or more bones. It may occur in any bone but the long bones; skull and ribs are most often affected. It is usually an incidental imaging finding. However, it may be complicated by pathological fracture and rarely by malignant change. It can also be associated with aneurysmal bone cysts [2]. In ~3% of cases, patients may also have endocrine diseases, skin pigmentation and precocious puberty; they together constitute McCune–Albright syndrome (MAS) [1, 2]. FD of ribs accounts for up to 30% of all benign chest wall tumours, and monostotic forms are about four times more common than polyostotic forms. It is typically present in the third or fourth decade of life as an asymptomatic mass.

CASE REPORT

A 22-year-old lady presented with cough and dyspnoea. She was diagnosed as a case of bronchial asthma. Her chest radiograph showed an incidental finding of a cystic mass lesion with calcified margins in the right upper zone extending outside the thoracic cage (Figs 1 and 2). A computed tomography (CT) scan showed a large well-defined expansile lytic lesion with a heterogenous pattern of enhancement with ground-glass matrix and with thick sclerotic margin extending along the length of anterolateral aspect of the second rib on the right side (Fig. 3). Histologically, the lesion consisted of osseous and fibrous components. The osseous component consisted of disorganized irregular 'Chinese alphabet' spicules of woven bone separated by abundant fibrous stroma (Fig. 4). The fibrous component was composed of cytologically bland spindle cells with no atypia of stromal cells (Fig. 5). Her clinical examination did not reveal any limb deformity, scoliosis, pelvic obliquity, facial asymmetry or any skin pigmentation. Her menstrual history revealed

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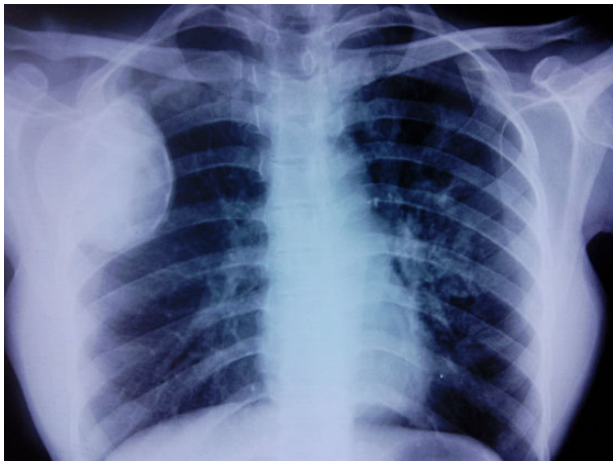

Figure 1: X-ray chest Posteroanterior view showing a cystic mass lesion in the right upper zone extending outside the thoracic cage.

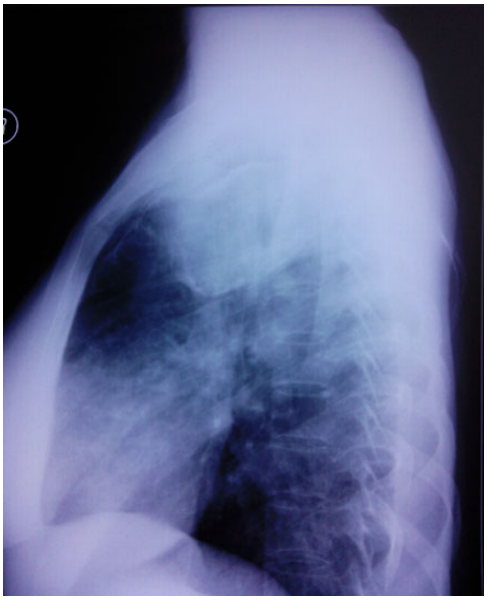

Figure 2: X-ray chest lateral view showing a cystic mass lesion in the right upper zone extending outside the thoracic cage.

menarche at the age of 14 year. Her thyroid functions were normal. Serum parathyroid hormone levels (40 pg/ml) and serum calcium levels (8.8 mg/dl) were also in normal range. Serum and urinary cortisol levels (12 μg/dl and 45 μg/24 h, respectively) were normal. Serum phosphate levels (1.2 mg/dl) were lowered. Growth hormone (GH; 10 ng/ml) and prolactin levels (18 ng/ml) were normal. Alkaline phosphate (189 U/l), urinary levels of *N*-telopeptide of collagen (43 BCE/nM creatinine), pyridinium cross-links (123 nmol/mmol creatinine) and deoxypyridinoline cross-links (34 nmol/mmol creatinine) were all elevated. Based on radiological and histological findings, final diagnosis was FD of rib. Surgery was advised for curative intention. Resection of the involved rib was performed with 1 cm clear margins. Histopathology of the lesion revealed no malignant change. There was no recurrence of the disease after 6 months of follow-up.

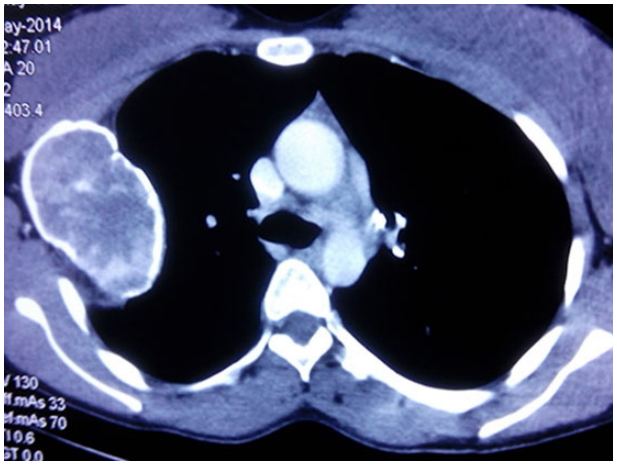

Figure 3: CT scan showing a well-defined expansile lytic lesion with a heterogenous pattern of enhancement with ground-glass matrix and with a thick sclerotic margin extending along the length of anterolateral aspect of the second rib on the right side.

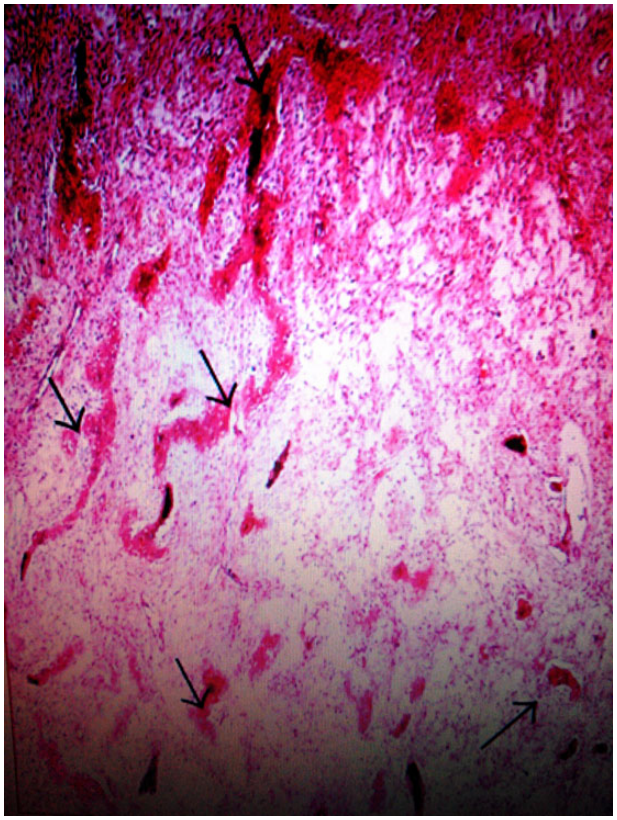

Figure 4: Histology of lesion showing osseous and fibrous components. The osseous component consists of disorganized irregular 'Chinese alphabet' spicules of woven bone separated by abundant fibrous stroma.

DISCUSSION

FD is characterized by the progressive replacement of normal bone elements by fibrous tissue and immature woven bone. Both monostotic and polyostotic FD are non-neoplastic processes associated with postzygotic-activating mutations of signal-transducing G proteins encoded by GNAS1 on chromosome

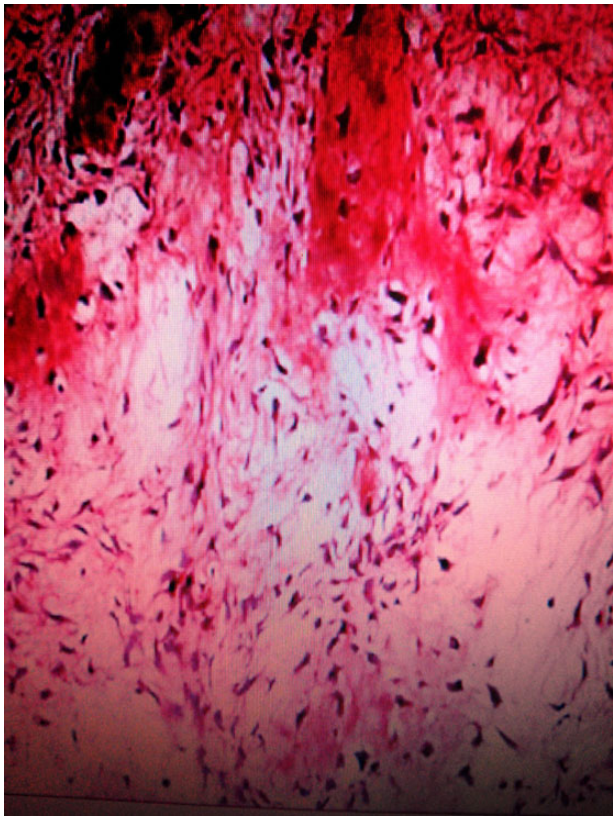

Figure 5: Histology showing the fibrous component composed of cytologically bland spindle cells with no atypia of stromal cells.

20 resulting in upregulation of cyclic adenosine monophosphate (cAMP) [3]. Osteoblasts carrying this mutation show increased proliferation and inappropriate differentiation, which results in fibrotic bone matrix [3, 4]. In its polyostotic form, FD may be associated with MAS (polyostotic FD, café-au-lait spots and endocrine dysfunction) and Mazabraud's syndrome (polyostotic FD and soft-tissue myxomas) [5]. Monostotic FD is generally asymptomatic and usually do not cause significant deformity. Monostotic lesions do not increase in size over the time, and the disease becomes inactive at puberty [2]. The most common sites of involvement include the ribs (28%), proximal femur (23%) and craniofacial bones (20%).

The imaging features of FD depend on the underlying histopathology of a given lesion. In the plain radiographs and CT, the lesions are classically described by their ground-glass appearance characterized by a variable degree of mineralization with a faint homogeneous increase in density. Radiographs show unilateral fusiform enlargement of medulla, deformity with cortical thickening and increased trabeculation. A characteristic 'ground-glass' appearance is created by the mixture of woven bone and fibrous components that replace the medullary space. More radiolucent lesions are composed of predominantly fibrous elements, whereas more radio-opaque lesions contain a greater proportion of woven bone [4]. Amorphous or irregular calcification is often seen in the lesion on CT scans. Bone scintigraphy is sensitive in the identification of the extent of skeletal FD, particularly in a polyostotic form [3, 6]. The differential diagnosis might include ossifying fibroma, osteoma, bone cyst, giant cell tumour or a malignancy of osseous origin [1, 3]. Malignant degeneration has been reported to occur in monostotic FD in 0.5% of patients [7].

Histopathologically, the lesion appears well circumscribed and sharply delineated by the host lamellar bone. It is composed of uniformly cellular fibrous tissue containing a proliferation of bland and uniformly spindle cells with sparse mitotic activity. Scattered across the fibrous matrix are lamellae or rounded nests of woven bone without significant osteoblastic rimming. There is some morphological variability in the woven bone spicules. The classic, most commonly seen pattern is that of curvilinear, 'Chinese alphabet' spicules of woven bone separated by abundant fibrous stroma. Less commonly, the woven bone may be deposited either in sclerotic, interconnected lamellae, cementoid bodies, or in orderly and parallel spicules [2–4].

Endocrine dysfunction may impact on the disease in different ways, and it depends on the endocrine disease and the degree to which the endocrine dysfunction is controlled. Precocious puberty, together with bowing, can lead to extremely short stature. Yet, when GH excess accompanies precocious puberty, stature can be normal, or even exceed the predicted height [8]. Renal phosphate wasting and hypophosphataemia, due to overproduction of the phosphate and vitamin D-regulating hormone, FGF-23, have been associated with earlier and more fractures. It is presumably by the direct effect of hypophosphataemia on inhibition of mineralization in FD, resulting in lesional osteomalacia [9]. In FD, disease extent is best assessed by bone scan, but activity can also be assessed by measuring serum and urine markers of bone metabolism. These include the bone formation markers including alkaline phosphatase, bone-specific alkaline phosphatase, osteocalcin, etc., and serum and urine markers for bone resorption *N*-telopeptide of collagen, pyridinium cross-links and deoxypyridinoline cross-links, etc. In general, all markers of bone metabolism are elevated in FD in parallel, relative to disease activity, and no specific assay is superior to another [10]. The severity of the outcome is proportional to the extent of the bone disease and association with other pathology—MAS. In patients with monostotic disease, the long-term prognosis is excellent. Treatment of FD is not codified and, generally, asymptomatic and stable lesions should simply be monitored. Surgery is indicated for confirmatory biopsy, correction of deformity and prevention of pathological and/or eradication of symptomatic lesions. When surgery is not possible, and in the polyostotic form, bisphosphonate therapy is indicated. Although FD is a disease of the osteoblast, bisphosphonates, which inhibit osteoclasts, are advocated for two reasons. First, it is felt that lesion expansion is mediated by osteoclastic resorption of adjacent normal bone, and that bisphosphonates inhibit this, and thus stop lesion expansion. Secondly, FD is a 'high-turnover' bone disease, sometimes with dramatic elevations in markers of bone turnover and occasionally evidence of increased numbers of osteoclasts in FD lesions.

We conclude that FD of rib may present as a mass in the lung. Knowledge of the various appearances, complications and associations of FD is important to ensure the accurate diagnosis and appropriate management. We believe that excision of involved rib should be indicated for both curative and diagnosis intention and to rule out malignancy.

CONFLICT OF INTEREST STATEMENT

None declared.

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