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Case report

Osteochondromyxoma; occurring in an adult at an unusual location

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Abstract

Osteochondromyxoma (OCHM) is an extremely rare benign, sometimes locally aggressive chondroid and osteoid matrix–producing tumour with extensive myxoid changes. It is seen in approximately 1% of patients with Carney complex (CNC) and constitutes one of its 11 diagnostic criteria. We report a case of a 36-year-old man who presented with an incidentally detected chest wall mass. Computed tomography (CT) suggested an osteochondroma. Intraoperatively, a globular, fragile lesion was identified arising from the posterior aspect of the seventh rib, and piecemeal excision was performed. Microscopically, the fragments showed a thick fibrous capsule enclosing mature bony trabeculae and hypocellular marrow spaces, with areas of fibro-myxoid stroma containing short plump to oval spindle cells with bland nuclei, with no atypia, mitoses, or necrosis. Systemic evaluation revealed no spotty skin pigmentation, breast lumps, or thyroid enlargement. Ultrasound of the thyroid and testes, echocardiogram, and adrenal imaging were unremarkable. Endocrine evaluation including serum cortisol, ACTH, and growth hormone levels was normal, excluding syndromic association. Although *PRKAR1A* mutation analysis, a supplementary criterion for CNC, was suggested, it was not performed. OCHM is an exceedingly rare benign bone tumour with an excellent prognosis following complete excision. Considering the patient's age and absence of clinical or endocrine features of CNC, this case most likely represents an isolated OCHM. However, long-term multidisciplinary follow-up is essential, as patients may develop diagnostic criteria for CNC in the future.
Key words: osteochondromyxoma, Carney complex, rib

Introduction

Osteochondromyxoma (OCHM) is an extremely rare benign, sometimes locally aggressive, chondroid and osteoid matrix producing tumour with extensive myxoid changes (1). It is seen in 1% of patients with Carney complex (CNC) and constitutes one of its 11 diagnostic criteria (2). It usually occurs in early life. However, it can occur in adults as an isolated entity (3). These tumours are commonly found in the nasal region and the diaphysis of the tibia and radius. Only a few cases have been reported in the rib, chest wall and spine. (1 – 4). We report a case of OCHM occurring in an unusual location.

Case History

A 36-year-old man presented with an incidentally detected chest wall mass. Computed tomography (CT) raised the

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possibility of an osteochondroma. Intraoperatively, a globular fragile lesion was identified arising from posterior aspect of the seventh rib. Piecemeal excision was performed. Multiple fragments of mucoid to hard tissue were received for histology. Microscopically, several fragments exhibited a thick fibrous capsule enclosing central areas of mature bony trabeculae and hypocellular marrow spaces (Figure 1). A few fragments with fibro-myxoid stroma, containing short plump to oval spindly cells with bland nuclei were present. No overt cartilaginous or chondroid differentiation was identified. There was no evidence of nuclear atypia, mitoses or necrosis. (Figure 2). The diagnosis of OCHM was made.

The patient was followed up in the orthopaedic clinic after surgical excision. On general physical examination, he appeared healthy and asymptomatic. There were no features suggestive of CNC—specifically, there was no spotty skin pigmentation, lentigines, or other pigmented cutaneous lesions. Breast examination was unremarkable, with no palpable masses or gynecomastia. There was no thyroid enlargement. Echocardiogram was normal excluding cardiac myxomas.

Given the known association of OSHM with CNC, a comprehensive endocrine evaluation was undertaken to exclude related systemic manifestations.

Endocrine assessment was normal. Bilateral adrenal imaging (Computed tomography) was normal, with no nodularity or enlargement. Serum cortisol and plasma ACTH levels were within normal limits, thereby excluding

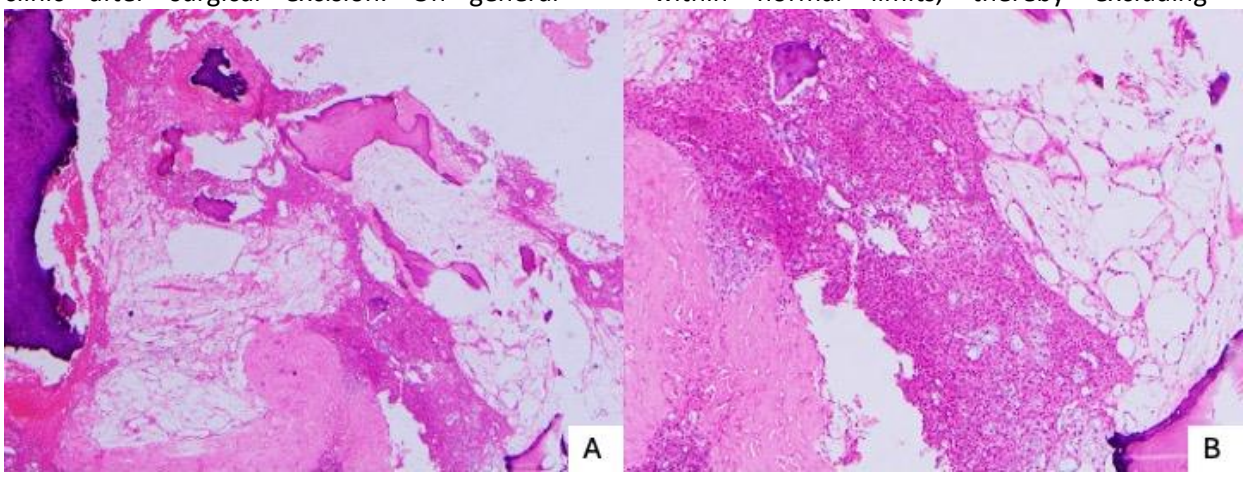

Figure 1: (A) Mature bony trabeculae with hypocellular marrow spaces and fibro-myxoid stroma (Haematoxylin and eosin (H&E) x40); B) Myxoid stroma containing bland spindle cells (H&Ex100)

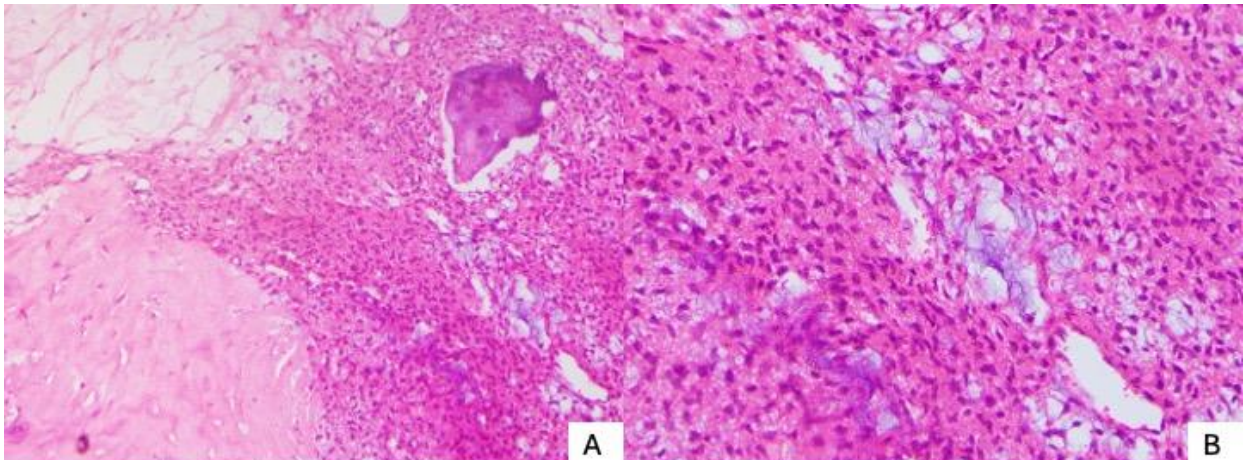

Figure 2: Bland spindle cells in the myxoid stroma (H&Ex200) B) Short plump to oval spindle cells with bland nuclei, with no atypia, mitoses, or necrosis (H&Ex400)

primary pigmented nodular adrenocortical disease (PPNAD) and other causes of endogenous hypercortisolism. Growth hormone levels were normal, ruling out acromegaly. Thyroid function tests (TSH, free T4, and T3) and thyroid ultrasonography were normal, excluding thyroid involvement. There was no testicular enlargement and ultrasound scan of testes revealed no evidence of a neoplasm.

Discussion

The diagnosis in the present case was established based on morphological features. Although no overt cartilaginous differentiation was identified, OCHM is known to exhibit marked histological heterogeneity, with variable proportions of myxoid, osteoid, fibrous, and chondroid components. Prior studies have emphasized that the relative representation of these elements may vary considerably both within individual tumours and between cases, and that chondroid differentiation may be minimal or even absent in some lesions (5, 6). Cellularity is typically inversely proportional to the amount of extracellular matrix, accounting for the predominantly hypocellular appearance, although some cases may show relatively hypercellular areas (5, 6). Awareness of this broad morphological spectrum is essential to avoid underdiagnosis or misclassification, particularly when one or more of the expected components is inconspicuous or absent. Therefore, despite the absence of a definite cartilaginous component, the constellation of prominent fibro-myxoid stroma, bland spindle cells, and osteoid/bony trabeculae with hypocellular marrow spaces supports the diagnosis of OCHM.

Carney complex (CNC) is a rare autosomal dominant multiple neoplasia syndrome associated primarily with inactivating mutations of the *PRKAR1A* gene located on chromosome 17q22–24, designated as CNC1. This gene encodes the regulatory subunit type I alpha of protein kinase A (PRKAR1A), which plays a crucial role in the cyclic adenosine monophosphate (cAMP) signalling pathway. Mutations lead to dysregulation of this pathway, resulting in tumorigenesis in multiple endocrine and non-endocrine tissues. Another locus, CNC2, mapped to chromosome 2p16, has also been implicated; however, its precise function remains to be elucidated (7).

The diagnosis of Carney complex is established when a patient fulfils two or more major diagnostic criteria, or alternatively, one major criterion together with one supplemental criterion (7).

The major diagnostic criteria for CNC include: 1) spotty skin pigmentation with a characteristic distribution (involving lips, conjunctiva, and genital or mucosal surfaces); 2) cutaneous, mucosal, or cardiac myxomas; 3) breast myxomatosis or characteristic magnetic resonance imaging features; 4) primary pigmented nodular adrenocortical disease (PPNAD); 5) acromegaly due to a growth hormone–producing pituitary adenoma; 6) Large-cell calcifying Sertoli cell tumor (LCCSCT) or characteristic testicular calcification; 7) thyroid carcinoma or multiple hypoechoic nodules on thyroid imaging in a prepubertal child; 8) psammomatous melanotic schwannoma; 9) multiple blue nevi or epithelioid blue nevi; 10) multiple breast ductal adenomas; and 11) osteochondromyxoma. The supplemental diagnostic criteria comprise 1) an affected first-degree relative with Carney complex; 2) identification of a pathogenic variant in PRKAR1A or PRKACB, and 3) an inactivating mutation of the PRKAR1A gene (7).

In the present case, apart from the histologically confirmed OCHM, the patient did not exhibit any other major clinical criteria of CNC. There were no cutaneous pigmentary lesions, cardiac or cutaneous myxomas, breast lumps or endocrine abnormalities suggestive of systemic involvement. In addition, there was no family history of CNC or related clinical features among first-degree relatives. Molecular genetic analysis for *PRKAR1A* or *PRKACB* mutations was suggested to further evaluate for a possible syndromic association; however, this testing was not undertaken due to resource limitations.

OCHM is an exceptionally rare manifestation of Carney complex, reported in only 1% of affected patients (2, 4, 8), and it usually presents early in life (3, 9), often during childhood or adolescence. Considering the age of 36 years at presentation and the absence of any clinical or familial features of CNC, this lesion most likely represents an isolated OCHM (3). Alahmhari et al. reported a nasal OCHM occurring in a 14-year-old girl with similar histological features, described as an isolated OSCM (4). However, a multidisciplinary approach and regular long-term follow-up are warranted to monitor for potential recurrence or late development of syndromic features.

Complete surgical excision is the definitive management for OCHM, as incomplete removal may lead to local recurrence. The prognosis following complete resection is generally favourable, with no reported instances of malignant transformation or metastasis in the literature (4, 7, 10).

On CT imaging, the lesion was initially interpreted as an osteochondroma, a conclusion possibly influenced by its atypical rib location and the exceptional rarity of OCHM. However, the absence of definite cortical and medullary continuity with the underlying bone, a hallmark feature distinguishing OCHM from osteochondroma, serves as an important diagnostic clue, although this may not always be readily appreciable in small lesions (2, 7). The overlapping radiological features between these entities, especially in an atypical location such as the rib, further highlight the diagnostic challenge and emphasize the importance of histopathological evaluation for definitive diagnosis.

Conclusion

OCHM is an exceedingly rare benign bone tumor with an excellent prognosis following complete surgical excision. Although it constitutes one of the major diagnostic criteria of CNC, it is seen in only about 1% of patients with CNC. Histopathological examination remains the definitive method for diagnosis, particularly in atypical locations or when radiological findings are inconclusive. In this case, the absence of syndromic or endocrine features, the atypical rib location, and the age of the patient favour an isolated form of OCHM. However, long-term follow-up with a multidisciplinary approach is essential, as patients may meet the diagnostic criteria for Carney complex in the future.

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