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Malignant transformation of multiple exostosis: a case report

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Abstract

Introduction: Osteochondroma is a benign tumor of bone. Malignant transformation of Osteochondroma is the most devastating complication one can encounter. Osteochondroma can transform into any malignancy like Osteosarcoma, Chondrosarcoma and Ewing sarcoma. Malignant transformation is more common in patients with multiple exostosis. Recognition of this malignant transformation is needed to predict the patient's outcome.

Case presentation: A 26-year-old male patient came with complaints of a mass in the left knee region for the past 7 years. X-ray of the knee showed multiple pedunculated exostosis on either side of the distal end of the femur, tibia and fibula. Histopathological examination revealed a bony lesion with a cartilaginous cap of increased thickness and cellularity. The cartilaginous cap possesses plump chondrocytes showing binucleation-forming nodules with mild atypia. The cartilaginous cap undergoes endochondral ossification, suggesting the possibility of a secondary peripheral atypical cartilaginous tumor from osteochondroma of the tibia.

Discussion: Chondrosarcoma is a heterogeneous type of primary bone cartilaginous malignancy with variable clinical outcomes. Malignant transformation of osteochondroma in the appendicular skeleton was named atypical cartilaginous tumor; in the axial skeleton, it is named Grade 1 Chondrosarcoma.

Conclusion: Differentiation between osteochondroma and its malignant transformation can be possible if made in a multidisciplinary setting such as clinical history, radiological findings along with histology to confirm the diagnosis.

Keywords: Osteochondroma, Chondrosarcoma, Bone tumour

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Introduction

Osteochondroma the most common benign tumour of bone accounts for about 35% of benign bone tumours affecting 3% of the population (1-3). Osteochondroma arises from the metaphysis of bones most commonly in the second to third decade of life. Commonly affected bones are long bones of the leg, scapula and pelvis (4).

Osteochondroma usually presents as a painless, asymptomatic mass and is usually found as an incidental finding. Osteochondroma are benign cartilage forming tumor derived from aberrant cartilage of the perichondral ring that may present either as solitary osteochondroma or multiple hereditary exostosis leading to syndromic manifestation of the lesion (5,6).

Osteochondromas are mostly treated by surgical excision of the lesion either partially or completely. The most common complication of osteochondroma is its malignant transformation. Osteochondroma can transform into osteosarcoma, chondrosarcoma and Ewing sarcoma (7,8). Malignant transformation of osteochondroma into chondrosarcoma is considered as drastic complication of osteochondroma accounting for about less than 1% of the cases (9). 3-5% of patients with multiple osteochondromas undergo malignant transformation (10). Here we present a case of a young male with multiple exostosis presenting with malignant transformation.

Case presentation

A 26-year-old male patient came with complaints of pain in the left knee for the past 7 years. The patient took medications for 3-4 years as analgesics, but after 4yrs since patient was suffering from more pain and swelling over the knee joint, he took X-ray, X-ray showed mass in the left knee region. History of pain during rest and walking. The swelling was insidious in onset, progressive in nature, not mobile, hard in consistency, fixed to the underlying bone. No history of any previous surgery or chemo or radiotherapy.

The patient had undergone radiological examination and X-ray knee showed multiple pedunculated exostosis noted on either side of distal end of femur, Proximal end of tibia and fibula (Figure 1).

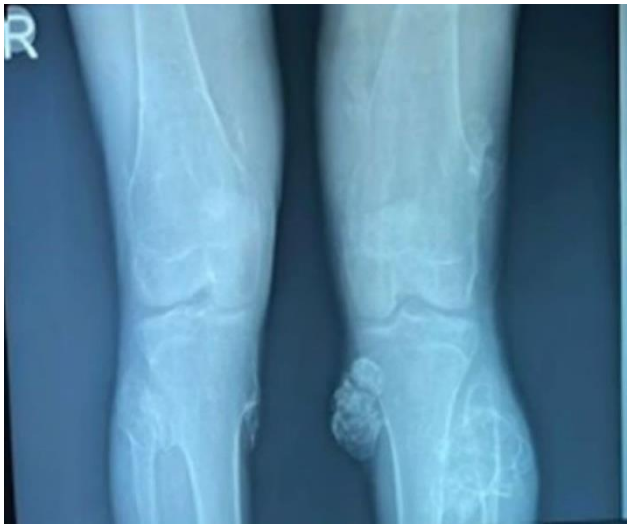

Figure 1. Xray image of the patient showing lobulated mass over tibia and fibula.

Reconstructed 3D imaging showed multiple sessile and pedunculated exostosis noted in multiple visualized bones, largest measuring 6.2 x 6.2 x 6.6cm. Pedunculated metaphyseal exostosis away from joint space in the medial aspect of proximal tibia with calcification of chondroid matrix, suggesting the possibility of Osteochondroma with sarcomatous transformation. (Figure 2). There is no significant family history of any bone lesions.

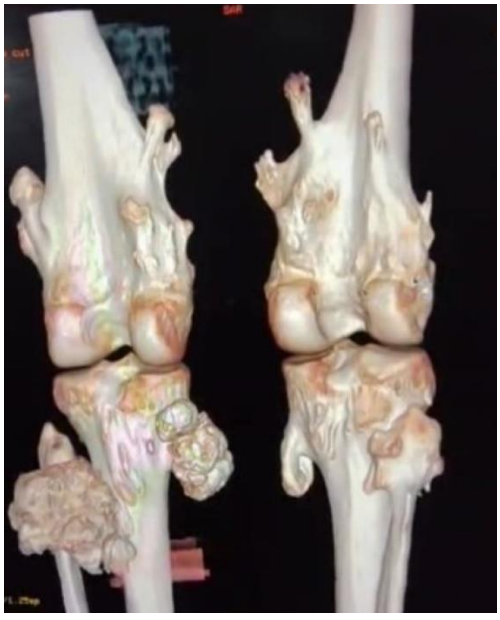

Figure 2. Reconstructed 3D image of the patient showing multiple pedunculated mass over the tibia and fibula.

Excision of a single pedunculated mass from the lateral aspect of tibia was received which showed a single bony tissue with a cartilaginous cap totally measuring

6 x 3.5 x 4 cm. Cut surface shows bone tissue measuring 3.5 x 2.5cm with irregular nodular cartilage cap of varying thickness measuring 2.5cm at its thickest portion permeating into the bony stalk (Figure 3).

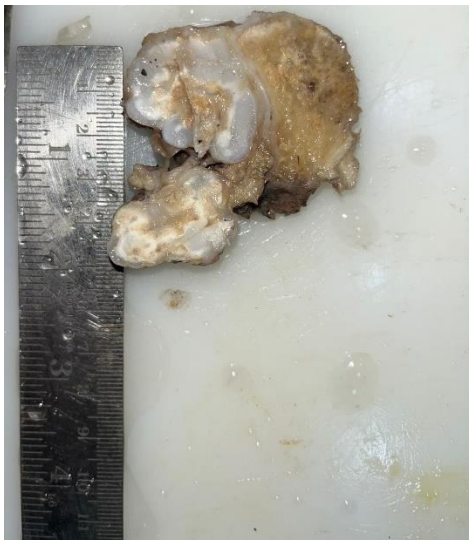

Figure 3. Bony stalk with cartilaginous cap of varying thickness and permeation into the bone.

Histopathological examination revealed a bony lesion with cartilaginous cap of increased thickness and cellularity. Cartilaginous cap increased cellularity possess plump chondrocytes showing binucleation forming nodules with mild nuclear enlargement, irregularity and atypia. Cartilaginous cap undergoes endochondral ossification as in a case of osteochondroma, suggesting the possibility of Secondary peripheral atypical cartilaginous tumor from osteochondroma of tibia (Figure 4,5).

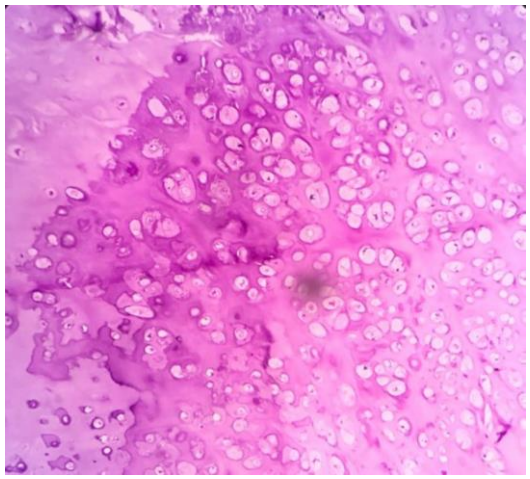

Figure 4. Histopathological image showing cartilage undergoing endochondral osssification (H&E stain, 10X).

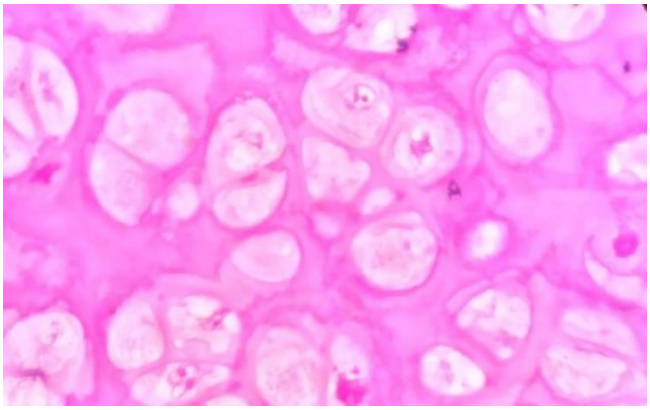

Figure 5. Histopathological image showing nodules of chondrocytes exhibiting mild atypia (H&E stain, 40X).

Discussion

Hereditary multiple osteochondromas (HMO), an autosomal dominant disorder involves two or more exostoses in the axial or appendicular skeleton. It is diagnosed by presence of two or more osteochondromas, detected radiographically in the metaphyseal ends of the long bones (11).

Chondrosarcoma is a heterogeneous type of primary bone cartilaginous malignancy with variable clinical outcomes (12). These are locally aggressive, hyaline cartilage-producing neoplasm arising within the cartilaginous cap of a pre-existing osteochondroma, tumours in the appendicular skeleton can be called as peripheral atypical cartilaginous tumor and tumours of the axial skeleton (including the pelvis, scapula, and skull base) can be called peripheral chondrosarcoma Grade 1 (13).

The incidence of chondrosarcoma varies between various bones with Ileum (19%), followed by the scapula (15%), pubic bone (10%), ribs (10%), tibia (12%) and femur (11%) (13).

Patients with multiple osteochondromas carrying germline mutations in EXT1 or EXT2 are at increased risk of developing ACT/CS1 within the cartilaginous cap of osteochondromas. Malignancy risk in case of multiple osteochondromas is as high as about 5% when compared to solitary osteochondromas which is about 1% (14).

Functional loss of genes EXT1 and EXT2 encoding glucosyltransferases which is involved in the synthesis of heparan sulfate causes Hereditary multiple exostosis

(HME). HME genetic transmission occurs in autosomal dominant pattern or loss of heterozygosity or haploinsuffiency or through mutations in post-transcriptional regualtory pathways.

Even isolated mutations of EXT1 and EXT2 gene cause pathology affecting the patient's growth. Malignant transformation is usually rare accounting for about 2 to 4% in patients affected by HME. A well-differentiated carcinoma is usually diagnosed, but very rarely osteosarcomas and dedifferentiated chondrosarcomas from bone could arise (15).

Differentiation between osteochondroma and its malignant transformation can be possible if made in a multidisciplinary setting such as clinical history, radiological findings along histology to confirm the diagnosis (14).

Treatment of Multiple exostosis is surgical removal, especially in symptomatic cases irritating adjacent structures. Though the treatment strategies are limited, precise diagnosis is essential for management. In Future, molecular analysis of EXT1 and EXT2 genes is essential for understanding the disease at molecular and cellular level and reveals new treatment options or therapeutic targets in both Hereditary multiple exostosis and chondrosarcoma (15).

Chemotherapy and radiation are not indicated for chondrosarcoma since they are resistant to both. Grade I chondrosarcoma with minimal rate of metastasis in the extremities are treated by intralesional curettage, high speed burring and adjuvant treatment with internal fixation and packing using phenol or ethanol or liquid nitrogen. Lesions in pelvis or axial skeleton needs wide local excision (15).

The 5-year and 10-year local recurrence rates for secondary peripheral chondrosarcoma are 15.9% and 17.5% respectively. The 5-year and 10-year mortality rates are 1.6% and 4.8% respectively. Local recurrences are possible due to incomplete excision in inaccessible locations (14).

Conclusion

This case report deals with the most common bone tumour osteochondroma undergoing malignnat transformation which emphasize the fact that multiple disciplinary evaluation, as well as careful gross examination, helps us to make the proper diagnosis at the appropriate time which helps in improving the prognosis and outcome of the patient.

We hope that this case report raises awareness among clinicians and pathologists to this possible transformation of osteochondroma to chondrosarcoma, and that thorough investigation drives further development in the diagnosis and safe treatment for improving patient outcomes.

Author contribution

BB: Conceptualization, Data curation, Formal analysis, Investigation, Methodology, Project administration, Resources, Software, Supervision, Validation, Visualization, Writing original draft. **SK**: Conceptualization, Data curation, Formal analysis, Funding acquisition, Investigation, Methodology, Project administration, Resources, Visualization, Writing original draft. **JS**: Conceptualization, Data curation, Formal analysis, Funding acquisition, Investigation, Methodology, Project administration, Resources, Software, Supervision, Validation, Visualization, Writing original draft, Writing review & editing

Conflict of interest

The authors declare that they have no competing interests.

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