

Rare Case of Primary Sarcomatous Transformation of Giant Cell Tumour

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Abstract: *Giant cell tumors of bone, also known as osteoclastomas, are relatively common bone tumors and are usually benign. Malignant transformation of GCT can either be primary or secondary, secondary being much more common than primary. Here we present a case of primary sarcomatous transformation of giant cell tumour of the distal humerus in a 42 year old man with probable lung and vertebral metastasis.*

Keywords: giant cell tumour, malignant, sarcomatous transformation, primary malignant transformation

1. Introduction

Giant cell tumor (GCT) is a primary locally aggressive bone neoplasm characterized by stromal mononucleated cells associated with uniformly distributed osteoclast-like giant multinucleated cells. This tumor type accounts for 4-5% of all primary bone tumors and 20% of benign bone tumors. Skeletally mature patients ranging from 20 to 45 years of age, especially women, are affected [1]. GCT usually involves the epiphyseal or meta-epiphyseal region of long bones, particularly the femur and tibia, and appears radiographically as a purely osteolytic eccentric lesion. Malignancy in GCT (MGCT) cases is defined as high-grade sarcomas originating in a GCT (primary) or at the location of a previous well-documented GCT (secondary). Primary malignancy in GCT (PMGCT) is the rarest type, and it seems to behave less aggressively than the secondary type does. [2] Here, we report the clinicopathological features of a rare case of PMGCT that was referred to the Department of Radiodiagnosis JNMC, Wardha for MRI elbow joint.

2. Case Report

42 years old male, pharmacist by profession presented with a swelling over the right elbow since 3 years. The swelling was associated with dragging type of pain and restriction of movements in the right elbow. He also complained of an exponential increase in size of the swelling since 1 year i.e. the swelling had become three times the size compared to an year ago. No h/o fever, no h/o trauma, no h/o previous surgery or radiation treatment. On clinical examination, the swelling was found to be mostly located in the posterior aspect of the right elbow, of variable consistency and non-tender on palpation. However there was local rise in temperature and restriction of flexion extension at elbow.

The patient had a previous X-Ray done 1 year ago when he underwent consultation for the first time. He was advised surgery but he deferred any treatment. The mentioned plain radiograph has been shown below. He underwent X-ray at our institute upon referral from Orthopaedic department which revealed a large expansile lytic lesion in the lower end of humerus in the epiphyseo-metaphyseal region with thinned out expanded cortex with areas of cortical breach and destruction. The lesion overall showed a soap bubble appearance with a solid Periosteal Reaction and narrow zone of transition. There was also e/o associated large soft tissue swelling around the elbow joint in relation with the lesion.

Compared to the X-ray which was done 1 year ago, there was pronounced destruction, cortical erosion and notable increase in the size of the soft tissue component of the mass.

CT scan of right elbow was performed and it confirmed the presence of the large expansile epiphyseo-metaphyseal lytic lesion arising from the distal humerus causing cortical expansion, erosion and destruction. Large Soft tissue Component with multiple hypodense areas within were also noted. It was established that the bony component of the mass extended only upto the articular surface of humerus and the Joint space and articulating surface of radius and ulna were normal. Findings which were noted in the X-Ray could be confirmed on CT scan apart from the ones mentioned.

Patient underwent MRI scan for the swelling for further characterization and it revealed a large well-defined irregularly marginated heterogeneous intensity lesion in the meta-epiphyseal region of the distal humerus reaching up to the articular surface of humerus with areas of cortical breach with extension into the surrounding soft tissue. The soft tissue component was extending across the elbow joint and involving the anterior and posterior compartment of the distal arm and the flexor and lateral compartments of the proximal forearm. The soft tissue mass was predominantly solid in nature with multiple cystic spaces. On T1WI, the lesion was of heterogeneous and intermediate signal intensity with cystic spaces appearing hypointense. T2WI showed predominantly hypointense signal intensity with multiple cystic areas appearing hyperintense, few showing fluid-fluid levels. On Diffusion weighted imaging, there was restricted diffusion seen in the solid component with ADC values suggesting a malignant nature of lesion. T1+C revealed intense post-contrast enhancement in the solid component.

Radiological Investigations pointed towards an aggressive bone tumour, probably even malignant and hence, even though asymptomatic patient underwent an HRCT Thorax and CECT abdomen to rule out any metastasis. Surprisingly we found the evidence of multiple well-defined tiny nodules in the right lower lobe. Though not showing any obvious signs of malignancy, considering the aggressive nature of the primary tumour, this was dealt with high index of suspicion and follow-up scan after 2 months was advised. CECT abdomen study was unremarkable except for a vertebral compression fracture of the D12 vertebra. Although there was no lytic/sclerotic nature to the fracture

and there was no pedicle or posterior element involvement, the nature of incidence i.e. without any history of trauma, metabolic diseases and relatively young age, this spontaneous vertebral compression fracture was also looked upon as a suspicious lesion.

FNAC was performed from the lesion in the elbow and revealed Multinucleated Giant Cells in a background of mononucleated stromal cells which was otherwise non-specific.

3. Discussion

A giant cell tumor (GCT), also known as an osteoclastoma, is a relatively common, generally benign but aggressive and destructive tumor of bone that typically affect females slightly more commonly than males within the ages of 20-40 years old, following skeletal maturity.^[1] The most common region of GCT occurrence is at the end of the long bones, especially the knee joint area. The distal femur and proximal tibia region account for more than 50% of GCT location^[3]. These typically benign tumors can interestingly metastasize to the lungs and is seen in up to 5% of cases. In 1-3% of giant cell tumor cases, "spontaneous transformation to a high-grade malignancy" can occur^[4]. Malignant transformation can be primary or secondary, but this distinction can be difficult to assess. A primary malignancy is extremely unusual in giant cell tumors however malignant transformation in secondary tumors (tumors that recur several years later in the same location as an initial benign giant cell tumor, following surgical or radiotherapy) is more common^[5]. The prognoses of malignant giant cell tumors are poor and are that of high grade sarcomas. The primary therapy of choice is surgery or curettage, depending on the local spread of the tumor. Primary malignancy in GCT (PMGCT) is rare and represents less than 1% of GCT^[6].

The features of both benign giant cell tumors and those with malignant transformation are very similar. Grossly, the tumors are typically soft, lobulated and hemorrhagic with erosion of the adjacent bone. Radiographically, giant cell

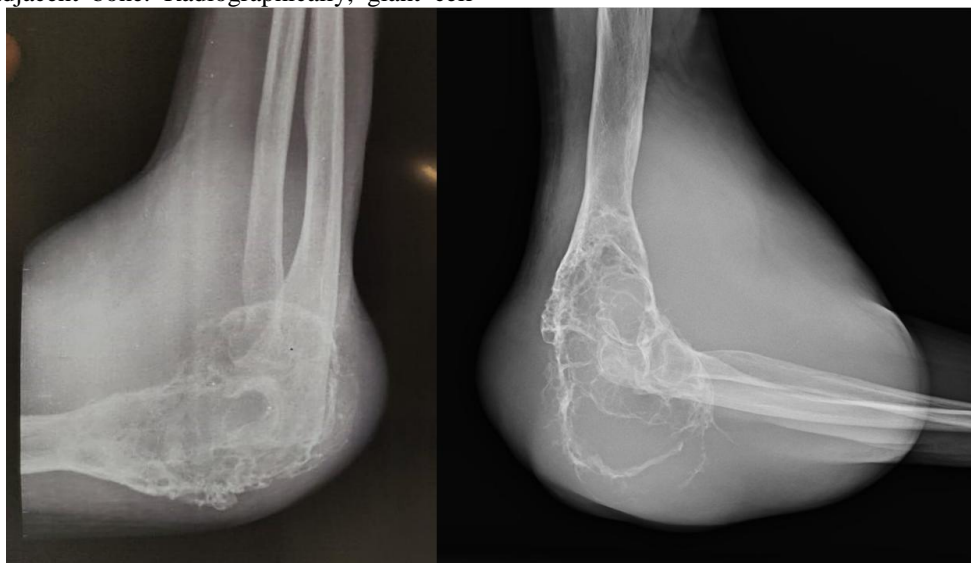
tumors are "aggressive osteolytic tumors with cortical destruction and soft tissue extension"^[7]

Histologically, there are sheets of ovoid or round mononuclear stromal cells interspersed with uniformly distributed, sparse, large, osteoclast-like giant cells with centrally located nuclei without atypia.^[8] The histopathological classification criteria for the sarcomatous component in PMGCT are not well established in the literature.

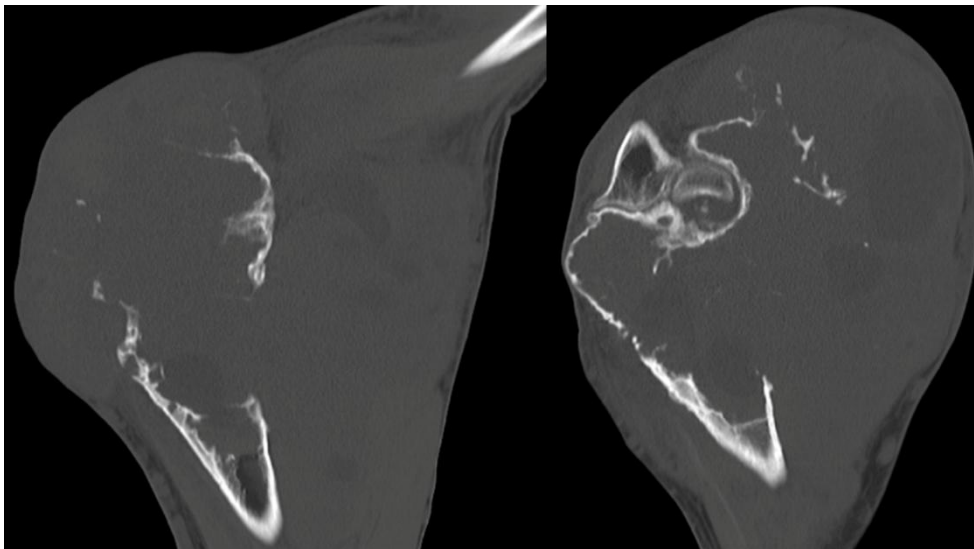
The diagnosis of a malignant GCT was made in our case because of the typical appearance of GCT i.e. a lytic expansile metaphyseal-epiphyseal lesion in a mature skeleton reaching upto articular surface with clear matrix and soft tissue component although distal humerus is an unlikely site for GCT. The destructive pattern, enhancing soft-tissue component, exponential increase in size, possibility of distant metastasis, malignant nature of lesion on DWI imaging, and the FNAC report of multinucleated giant cells (representative of osteoclasts) supported a malignant nature of the mass. The negative history for previous surgical or radiation treatment rules out a secondary malignant transformation. Hence, the final diagnosis of Primary Sarcomatous Transformation of Giant Cell Tumour was made.

4. Conclusion

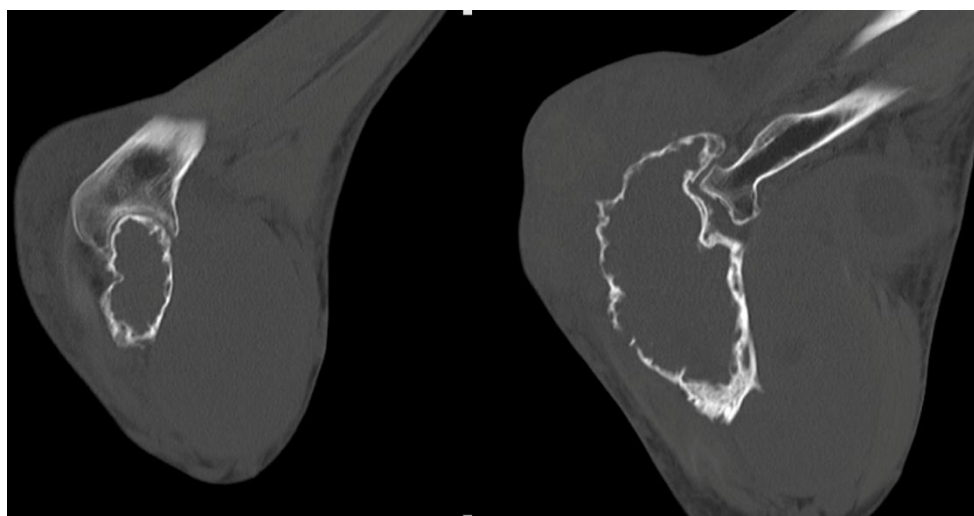
Giant cell tumors of bone are commonly benign but locally destructive tumors. Malignant transformation can be difficult to diagnose clinically, radiographically, and grossly due to the fact that the malignant components are interspaced within the benign components and can be easily missed on biopsy or when sections are taken at the grossing bench. Hence, even in the absence of malignant cells on FNAC, a large enhancing soft tissue component, sudden increase in size of the lesion and distant metastasis should arouse the suspicion of sarcomatous transformation even though extremely rare.



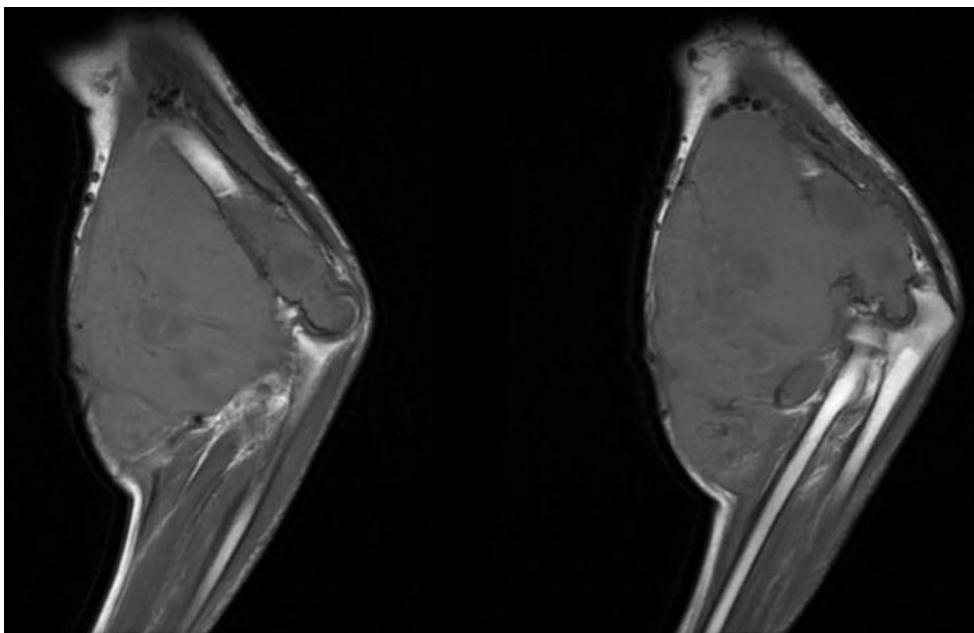
Lateral X-Ray right elbow: One year old and current X-rays showing the expansile lytic lesion of distal humerus.



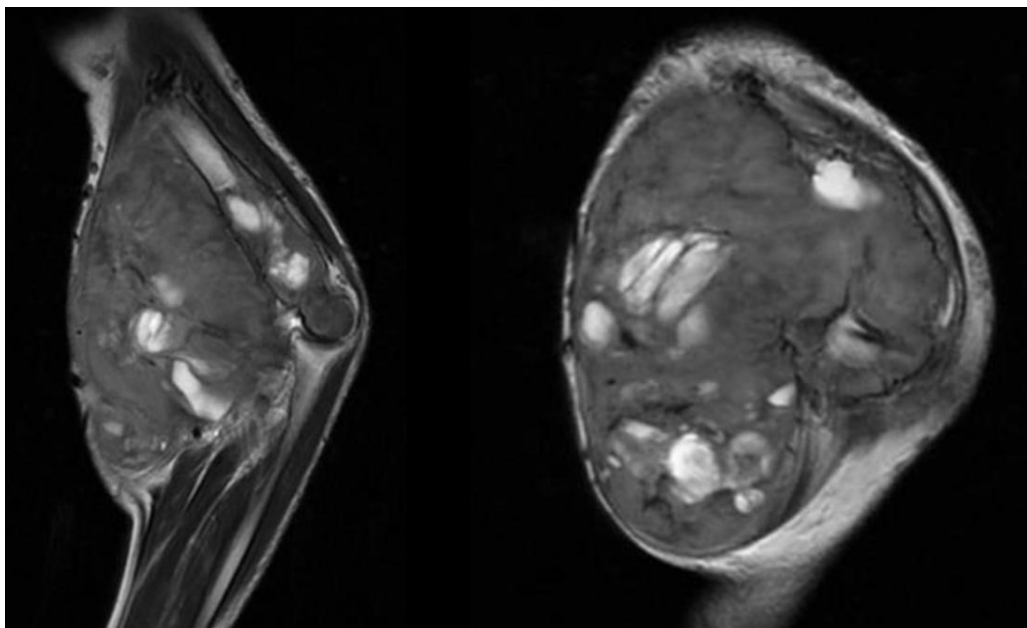
CT scan of right elbow showing the expansile lytic lesion of distal humerus with soft tissue component



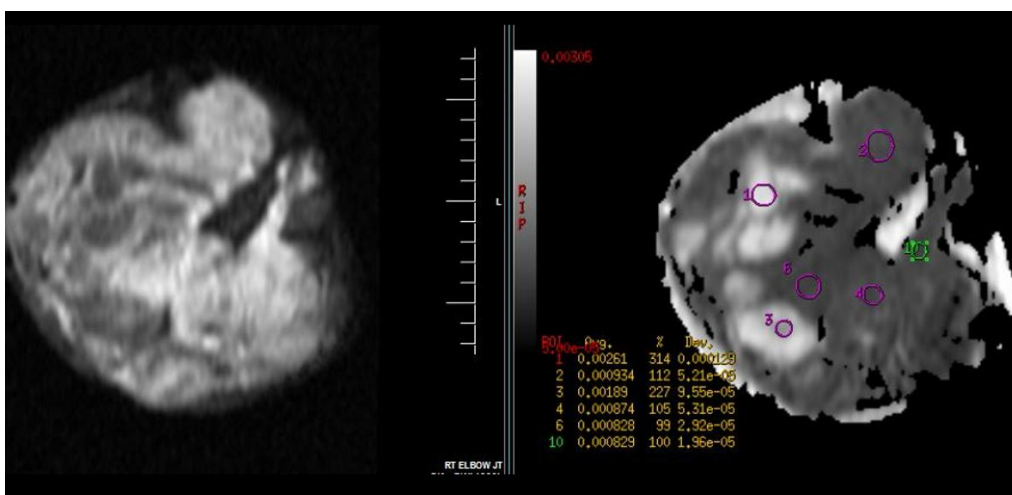
CT scan of right elbow showing the lesion extending up to articular surfaces of humerus with radius and ulna without involvement of the joint space and preserved radial and ulnar heads



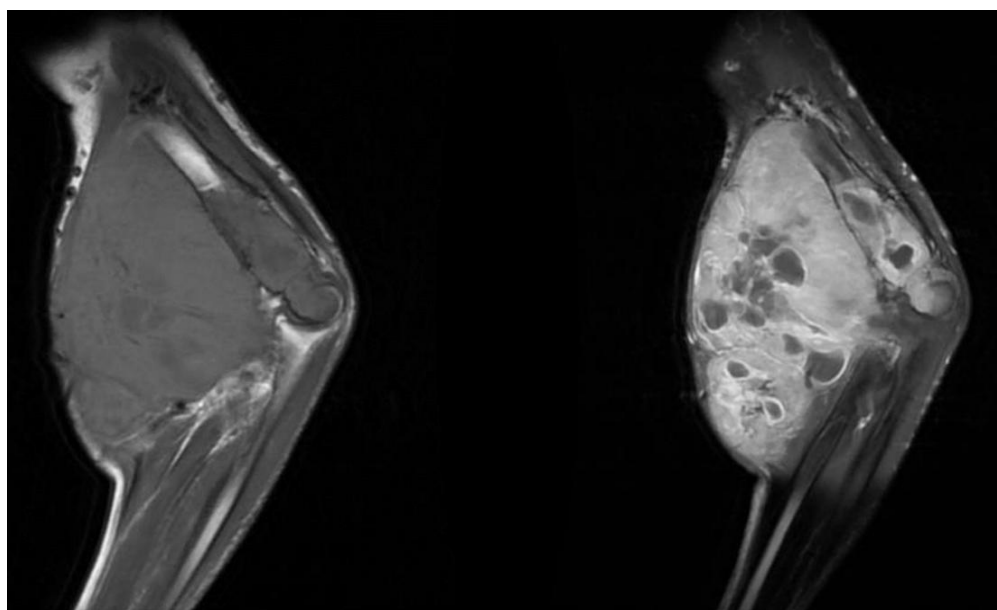
Sagittal T1WI of right elbow showing large soft tissue lesion which is in continuation with the bone extending upto articular surfaces of humerus with radius and ulna without involvement of the joint space and preserved radial and ulnar heads



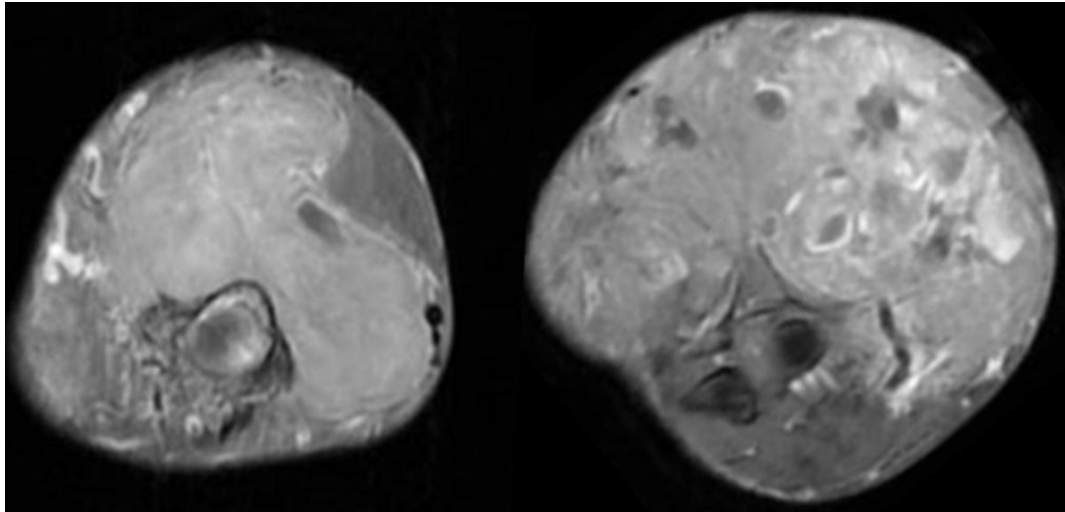
Sagittal and Axial T2WI showing heterogeneous nature of the mass with cystic components



DWI and corresponding ADC images showing restricted diffusion of the mass and ADC values <0.67 indicating a malignant nature



Sagittal T1W1 and Contrast enhanced T1 images showing vivid but heterogenous contrast enhancement



Axial T1+C images at the level of arm and forearm showing the extent of the enhancing soft tissue mass

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