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Case Report

Primary leiomyosarcoma of femur

Nishit Bhatnagar^{a,*}, Purushotham Lingaiah^a, Anurag Tiwari^a,
Nidhi Mahajan^b, Sumit Arora^c, Anil Dhal^d

^a Senior Resident, Department of Orthopaedic Surgery, Maulana Azad Medical College & Associated Lok Nayak Hospital, New Delhi 110002, India

^b Assistant Professor, Department of Pathology, Maulana Azad Medical College & Associated Lok Nayak Hospital, New Delhi 110002, India

^c Assistant Professor, Department of Orthopaedic Surgery, Maulana Azad Medical College & Associated Lok Nayak Hospital, New Delhi 110002, India

^d Director, Professor & Head, Department of Orthopaedic Surgery, Maulana Azad Medical College & Associated Lok Nayak Hospital, New Delhi 110002, India

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ABSTRACT

Primary leiomyosarcoma of the bone is exceedingly rare. In this case, we describe a middle-aged female with a primary leiomyosarcoma of the distal femur. The patient was treated by hip disarticulation. The patient continues to be disease-free at one-year follow-up.

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1. Introduction

Malignant tumours comprising of spindle-shaped cells with distinctive smooth muscle cell features are called leiomyosarcomas (LMS).¹ Most of these tumours originate in the uterus, gastrointestinal tract, and retroperitoneum.² Primary bony LMS is a rare clinical entity. Majority of the cases are secondary deposits from a distant source. LMS of the bone occurs more commonly in middle-aged males.² Long bones are particularly involved, distal femur being the most common site.³

The first description of a primary bone leiomyosarcoma was given in 1965.¹ Since then, to the best of our knowledge, around 90 such cases have been reported. We present the clinical features, radiological findings and pathological description of a primary leiomyosarcoma originating from the distal femur in a middle-aged lady.

2. Case report

A 50-year-old female of Indian subcontinent presented to our department with a history of pain and swelling around the left

* Corresponding author. Tel.: +91 9910465639.

E-mail address: nishitbhatnagar@yahoo.co.in (N. Bhatnagar).

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knee for the last six months. The patient had a history of insignificant trauma to the left knee following which she noticed pain that gradually increased in severity. There was an associated swelling on the anterior aspect of distal thigh that gradually progressed in size. Over the course of the last month, the patient had become unable to bear weight on the affected extremity. These symptoms were associated with history of loss of weight and appetite. There was no other contributory family and past history.

Examination revealed a globular ill-defined swelling on the anterior aspect of distal thigh, extending over to the knee joint, around 10 cm × 10 cm × 5 cm in size. The skin over the swelling was stretched and shiny with dilated tortuous veins coursing over the swelling. There was marked wasting of muscles of the thigh. The swelling was warm and tender on palpation. Consistency varied from bony hard to firm. The patient had a flexion contracture of 30° at the knee. Patellar mobility was also restricted. There was no neurovascular deficit.

Plain radiographs revealed a large expansile lytic lesion originating in the distal femur that predominantly involved the epiphyseal and metaphyseal regions. Superiorly, it also involved the adjacent part of distal femoral diaphysis. A pathological fracture was observed at the metaphysio-diaphysial junction of the distal femur. The lesion had permeative, ill-defined margins along with evidence of cortical thinning. Gross disruption of anteromedial cortex and breach

of the articular margin of the femur was noted with extension of the lesion into surrounding soft tissues (Fig. 1).

Multiplanar contrast-enhanced MR imaging was performed to obtain T1w, T2w and STIR images. The study revealed a large, well-margined mass lesion involving the epiphyseal and metaphyseal region of distal femur with cortical disruption and extension of the lesion into adjacent soft tissue. The lesion appeared hypointense on T1w-images and heterogeneously hyperintense on T2w and STIR images. Heterogeneous enhancement of the lesion was also noted in the post-gadolinium sequences. Anteriorly, the lesion was observed to be abutting the quadriceps tendon. Inferiorly, destruction of articular surface was observed with extension of mass into the knee joint. Posteriorly, the mass was seen to be in close proximity to the popliteal neurovascular bundle, but without any evidence of neurovascular invasion. Superior extension of the lesion was noted as hypointensity on T2w and STIR images, and post-contrast enhancement of the marrow in femoral diaphysis. Subcutaneous tissue and skin along the anteromedial aspect of thigh was suspected to be involved since it revealed mild heterogeneous enhancement in the post-gadolinium sequences (Fig. 2). Skeletal survey, chest X-ray, abdominal sonography and a CT chest and abdomen revealed no other site of primary or metastatic disease.

A core biopsy revealed loosely cohesive clusters of cells with oval elongated nucleus, moderate amount of cytoplasm

Fig. 1 – Plain radiographs showing an expansile lytic lesion with permeative ill-defined margins in the distal femur involving the epiphyseal and metaphyseal regions. A pathological fracture is seen at the metaphysio-diaphysial junction of the distal femur. Breach of the articular margin of the femur can be seen.

Fig. 2 – (A) T1w-images showing a hypointense mass lesion in distal femur abutting the quadriceps tendon and extending into the knee joint. (B) T2w images showing a heterogeneously hyperintense with extension of lesion into femoral diaphysis. (C) Post-gadolinium sequences showing heterogeneous enhancement of the lesion, subcutaneous tissue and skin along the anteromedial aspect of thigh.

and marked atypia which was suggestive of a high grade spindle cell sarcoma. Subsequently, a hip disarticulation was performed keeping in mind that the tumour was aggressive and extracompartmental.

The gross examination of amputated limb revealed grey white tumour mass 10 cm × 9 cm × 4 cm in size, originating in distal femur with gross cortical breach (Fig. 3a). Extra osseous extension to surrounding musculature and knee joint was noted. The histopathological examination of the amputated part showed a highly cellular tumour with spindle-shaped cells in sweeping fascicular sheets (Fig. 3b). The cells had an abundant eosinophilic cytoplasm containing elongated nuclei. There was moderate nuclear pleomorphism with many giant cells. Tumour cells were strongly positive for Vimentin and Smooth muscle actin (Fig. 3c). Immunohistochemistry for S-100 and CD 65 was negative. The resected margins were free of tumour tissue.

The amputation stump healed uneventfully. After one-year follow-up, there is no evidence of metastatic disease or local recurrence.

3. Discussion

Primary LMS of the bone accounts for less than 0.7% of all primary malignant bone tumours.⁴ It occurs most commonly in large bones, like distal femur and proximal tibia.⁵ Primary LMS is predominantly located in the metaphysis of long bones.⁶ Males and females are equally affected.⁷ Prognosis is based on the stage at time of presentation and histological grade.⁶

Radiologically, LMS are poorly defined, osteolytic lesions with a permeative or moth-eaten pattern of osseous destruction. Such a radiographic picture is not specific and can be mimicked by malignant fibrous histiocytoma, fibrosarcoma, chondrosarcoma, osteosarcomas and metastatic carcinomas. MRI is useful for delineating the intramedullary extent of tumour and its relationship to neighbouring soft tissue structures. Other imaging investigations like abdomino-pelvic ultrasonography and CT scan are useful in excluding the possibility of a metastatic disease.

Fig. 3 – (A) Gross specimen showing a heterogeneous tumour mass at the lower end of femur, with areas of haemorrhage and necrosis and breaching the cortex. (B) Scanner view showing a markedly cellular tumour with spindled out cells and dead necrotic bone. Inset – marked nuclear pleomorphism, tumour giant cells and mitotic figure (haematoxylin & eosin, 40×). (C) Tumour cells strongly express smooth muscle antigen (SMA) (Immunohistochemistry, 100×).

Since a primary LMS of the bone is exceedingly rare, we came up with the following differential diagnosis. A primary leiomyosarcoma of the gastrointestinal tract, uterus or elsewhere metastasizing to the distal femur was excluded by CT and ultrasonography. Low-grade intramedullary osteosarcoma was excluded due to lack of evidence of production of osteoid by tumour cells. Fibrosarcoma of bone, malignant fibrous histiocytoma and metastatic spindle cell carcinomas, though rare, were excluded by immunohistochemistry. The diagnosis of leiomyosarcoma is established by immunohistochemical demonstration of strong positivity for muscle markers like Vimentin and SMA.

Histologically osseous LMS can be classic type, epithelioid, myxoid and pleomorphic. It has been suggested that primary osseous LMS arises from vascular smooth muscle cells within bone.³ The alternate hypothesis suggests that it arises from a multipotent mesenchymal stem cell capable of smooth muscle differentiation.⁸

Primary osseous LMS is known to behave aggressively and is associated with poor prognosis. The principles of surgical treatment are similar to that of other primary malignant bone tumours. A wide resection should be done with a tumour free margin and an adequate cuff of normal tissue. Depending on the histological grade of the lesion and staging, a limb sparing resection with replacement of a

prosthetic device is recommended. Chemotherapy does not seem to provide an improved prognosis. The tumour also appears to be radioresistant.⁶

In our patient, the tumour was extracompartmental. It had caused a pathological fracture of the distal femur. Histopathological examination revealed it to be a high grade LMS. Taking these factors into account, we performed a hip disarticulation rather than an above knee amputation or limb salvage procedure.

We herein report the clinicoradiological and histopathological findings of a primary leiomyosarcoma of distal femur in a 50-year-old female patient.

Conflicts of interest

The authors have none to declare.

Ethical approval

A written informed consent was obtained from the patient authorizing radiological examination, photographic documentation, and surgery. He was also informed that the data concerning the case would be submitted for publication and he consented.

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