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ENIGMA IN DIAGNOSIS OF A MALIGNANT TUMOR IN THE TIBIA

AMRUTH
Department of Orthopaedic Surgery,COIMBATORE MEDICAL COLLEGE

Abstract : A 14 year old female patient presented to the orthopaedic department with complains of pain and swelling in the proximal 13rd of right leg since 2 months. History and clinical examination were indicative of a malignant tumour. The patient was investigated with radiography, histopathological examination, immunohistochemistry studies and bone scan to come to definite diagnosis. All the investigations and clinical tests were equivocal and could not provide a definite diagnosis. The definite diagnosis of synovial sarcoma was established only after radical excision of the tumour in the form of amputation. We present this case for the difficulty in establishing the diagnosis of synovial sarcoma and a review of the differential diagnosis considered.

Keyword :synovial sarcoma, osteosarcoma, ewings sarcoma, adamantinoma, metastasis

Introduction Synovial sarcoma is a mesenchymal spindle cell tumour which displays variable epithelial differentiation, including glandular formation and has a specific chromosomal translocation t(X;18) (p11;q11). The term synovial sarcoma is a misnomer since it doesn't arise from the synovium[1]. The term originates from the histological appearance of the cells, which can resemble synovial cells [1]. We report a case of synovial sarcoma which presented with varied clinical manifestations, radiological features and histopathological findings causing difficulty in diagnosis. Case report A 14 year old female patient Miss Muthumari presented with complaints of pain and swelling in the right leg just below the knee for 2 months with a recent increase in size of the swelling. There was a recent history of ulceration over the swelling with blood stained discharge from past 1 week. There was no history of fever, trauma, loss of weight or appetite. On general examination patient was moderately built and nourished. There were no signs of anemia. Patient had right inguinal lymphadenopathy, firm in consistency and tender on palpation. Patient was afebrile with stable vitals. Other systems examination were within normal limits. On local examination, a swelling measuring about 15X12 centimeters present over the proximal 1/3rd of right leg just below the knee joint. An ulcer measuring 2X2 centimeters present over the swelling with foul smelling blood stained discharge. Dialated veins present over the swelling. There were no scars. On palpation swelling was bony in consistency except at the center where it was soft. There was a local rise of temperature, tenderness present. The swelling was fixed to the underlying structures and to the overlying skin.

There were no similar swellings in the ipsilateral limb or elsewhere. There was no distal neurovascular deficit. A clinical diagnosis of osteosarcoma of right upper tibia was made and patient investigated. [Figure 1 and 2].

Figure 1 clinical picture showing the malignant lesion in right proximal tibia from lateral aspect

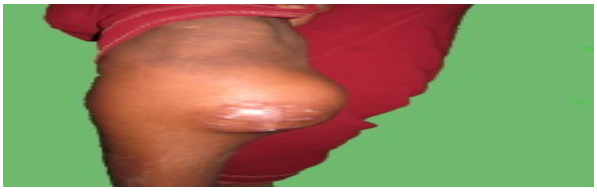

Figure 2 clinical picture showing the malignant lesion in right proximal tibia from front

Her hemoglobin was 12g/dl, total leukocyte count was 6,400 cells/cumm, with polymorphs 78% and lymphocytes 22%.The erythrocyte sedimentation rate was 10mm/hr. The renal function tests were within normal limits. The chest X-RAY was normal. Serum calcium was 10mg/dl and serum alkaline phosphatase was 50IU/L. X RAY right leg with knee AP and lateral views showed cystic lesions with intervening septae, expansion of the cortex and erosion of the medial and posterior cortex in the right upper tibia involving the diaphysis and metaphysis suggestive of ?Adamantinoma, ?Ewing's sarcoma.[figure 3]

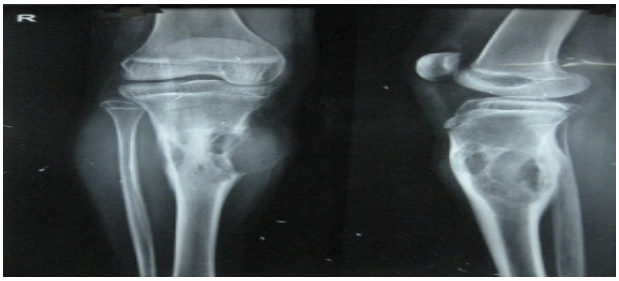

Figure 3 X RAY right leg with knee AP and lateral views showing Osteolytic lesion in the proximal tibia

The MRI of right leg was done which showed a hyper dense lesion in the diaphyseo-metaphysial region of right tibia with cortical destruction, invasion into the soft tissue and no neurovascular invasion which were suggestive of Ewing's sarcoma. [figure 4]

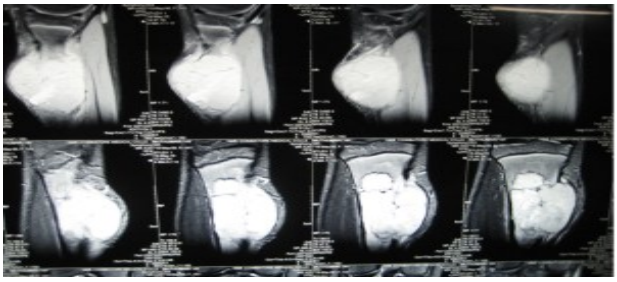

Figure 4 MRI of right proximal leg showing hyper dense lesion in the diaphyseo-metaphyseal region with cortical destruction and soft tissue invasion in the proximal tibia

Since the diagnosis was inconclusive open biopsy of the lesion was carried out which showed medium sized epithelial like cells arranged in sheets and nests invading the bone and stroma. Multinucleated giant cells seen-suggestive of metastatic deposit from poorly differentiated carcinoma.[figure 5]

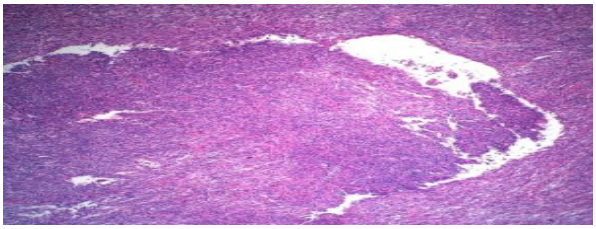

Figure 5 Histopathology of the open biopsy specimen showing medium sized epithelial like cells arranged in sheets and nests invading the bone and stroma

The immunohistochemistry study of the specimen showed that the tumor is strongly positive for cytokeratin, weakly positive for vimentin,CD 99, desmin and S-100 negative. These features suggested that the tumor is of epithelial origin(epithelioma) or a metastatic carcinoma. The patient was surveyed for a primary tumor in abdomen, thorax and genitourinary tract. All the clinical examinations were normal. Sigmoidoscopy and upper GI endoscopy were also normal. Isotope bone scan was done to rule out any other bone tumor. The results showed increased uptake in the upper third of right tibia at the site of known pathology, no scintigraphic evidence of any other skeletal metastasis and no other bone lesion.[figure 6 and 7]

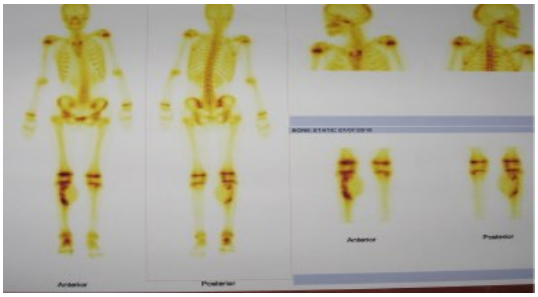

Figure 6 Isotope bone scan of the whole body showing increased uptake in the right proximal tibia

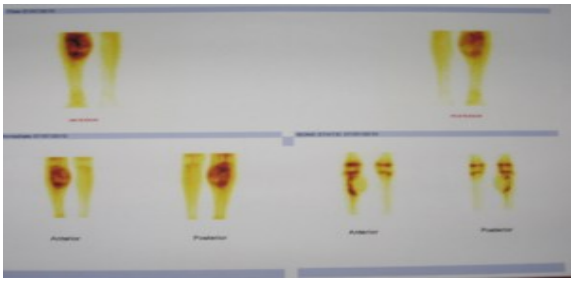

Figure 7 Isotope bone scan of the right leg showing increased uptake

Since the lesion started fungating, the patient had exquisite pain and the limb became non-salvageable an above knee amputation was planned and done. The specimen was sent forhistopathological examination. This specimen showed nests of epitheloid cells surrounded by spindlecells and a biphasic pattern which gave us the final diagnosis of synovial sarcoma.The patient was started on chemotherapy with Inj.Cisplatin 40mg in 500ml NS; iv infusion over 3 hrs.on days 1-3 with Inj.Doxorubicin 70mg in 500ml DNS; iv infusion on day 1 only.The amputation site healed completely following which patient was rehabilitated with suitableorthosis.Discussion:Our patient presented with a rapidly growing swelling in the proximal 1/3rd of right leg with ulceration and blood stained discharge. A clinical diagnosis of osteosarcoma was made and patient investigated with radiological and histopathological examination. Both these investigations were inconclusive.

Patient was also worked up for a metastatic deposit based on the immunohistochemistry report. Since no primary lesion was found out and the lesion was rapidly growing a definitive treatment in the form of above knee amputation was done and patient treated with chemotherapy. The differential diagnoses considered in this case are and differentiating points include [Table 1 and 2]

	AGE	SEX	BONE INVOLVED	SITE IN BONE
ADAMANTINOMA[3]	10-35YRS	-	TIBIA	DIAPHYSIO-METAPHYSEAL
EWINGS SARCOMA[2]	4-25YRS	MALE	LONG BONES	DIAPHYSIS
METASTATIC DEPOSIT[2]	>45YRS	MALE	AXIAL SKELETON UNCOMMON BELOW KNEES	METAPHYSIS
SYNOVIAL SARCOMA[3]	YOUNG ADULTS,RARE AFTER 40 YRS	-	KNEE	METAPHYSIO-DIAPHYSEAL
OSTEOSARCOMA[2]	10-20 YRS	MALE	LONG BONES	METAPHYSIS

Table 1 showing predominant age, sex, bone involved and the site in the bone in differential diagnosis namely adamantinoma, Ewing's sarcoma, metastatic deposit, synovial sarcoma and Osteosarcoma

Table 2 showing X ray and histopathological findings in the differential diagnosis namely adamantinoma, Ewing's sarcoma, metastatic deposit, synovial sarcoma and Osteosarcoma

Review of literature:

Synovial sarcoma is a mesenchymal spindle cell tumour which displays variable epithelial differentiation, including glandular formation and has a specific chromosomal translocation t(X;18) (p11;q11) [5]

It accounts for 2.5 to 10.5 per cent of all soft tissue sarcomas [6] .It has been reported from birth to 89 years but more common in young adults. Males affected more than females [2] The most common presentation is a swelling or mass with or without pain or t enderness. Pain or tenderness can be present for several years even though a mass could not be felt [2] There are no specific pre disposing factors. Occurrence has been associated with metal im plant used in hip replacement, and radiotherapy in Hodgkin disease. Specific chromosomal translocation that is presumably relevant in pathogenesis has been found [5] The most effective way to diagnose this cancer is by having a tissue biopsy [4]. Magnetic Resonance Imaging (MRI) is very useful in determining the extent to which the cancer has grown [3]. The radiographs might reveal a soft tissue mass of moderate density in close proximity to a joint. Occasional bone invasion can be seen. It may cause a periosteal reaction and multiple spotty calcifications (15 to 20%). Metastases to lungs and bone [3]. Histologically there are 3 types of synovial sarcoma [7, 8] Ø Biphasic : Both epithelial and spindle cell components. Ø Mono phasic : · Spindle cell · purely glandular · Ø Epithelial cell Poorly differentiatedImmunophenotyping [8,9]

Ø Cytokeratin : · 90% of Synovial sarcoma express cytokeratin in epithelial component. Ø Epithelial membrane antigen can be positiveØ Others: S 100(30%); CD99 (62%) Ø Muscle marker: Calponin , desmin , actin, Vimentint(X;18)(p11;q11) is the cytogenetic hallmark of synovial sarcoma, being present in more than 90% ofthe cases[5]

Synovial sarcomas may be treated by Surgery, Radiation Chemotherapy Complete surgical excision of the tumour, nearby muscle and lymph nodes is the best way of treating this cancer. Depending on the location and size of the tumour, it may be necessary to remove all or part of a limb [2, 9] Radiation is often used in conjunction with surgery to kill cancer cells. It can be given before surgery in order to shrink a tumour or afterwards to kill any remaining cancer cells. Chemotherapy has been proven highly effective with treating this form of cancer [8, 9] About 50% of synovial sarcoma recur, usually within 2 yrs., but sometimes up to 30 yrs. after diagnosis. 40% cases metastasize, commonly to lungs and bones and also in regional lymph nodes. Five years survival is 36-76% and 10 yrs. survival is 20-63% [8]. Conclusion: The diagnosis of synovial sarcoma at times can be very difficult. The aim of our presentation is to inform that synovial sarcoma should be considered as one of the differential diagnosis during examination of any swelling with features of malignancy.

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