Case Report

Jaffe–Campanacci syndrome: Any role for ^{99m}Tc-methylene diphosphonate bone and ^{99m}Tc-octreotide scans for evaluation of the disorder?

ABSTRACT

Jaffe–Campanacci syndrome (JCS) is a rare clinical disorder with almost unknown etiology. The main feature of this syndrome is skeletal involvement as nonossifying fibroma which may cause severe morbidity to these patients. X-ray imaging is the widely available modality for evaluation of skeleton, but radionuclide imaging modalities may have a role in workup. Herein, we present a case of JCS evaluated with ^{99m}Tc-methylene diphosphonate bone and ^{99m}Tc-octreotide scans for the extent of skeletal involvement. To the best of our knowledge, from over than 30 cases reported in the literature, no evaluation with radionuclide imaging has been done.

Keywords: Bone scan, Jaffe–Campanacci syndrome, octreotide scan

INTRODUCTION

Jaffe–Campanacci syndrome (JCS) is a rare condition with various clinical pictures including bony lesions as nonossifying fibroma (NOF), skin pigmentation, and other rarer abnormalities. The exact etiology has not been completely understood.[1,2] Skeletal involvement is the most prominent and debilitating feature of this syndrome, and pathologic fracture is a common initial presentation.[3] Although X-ray seems to be the modality of choice for diagnosis of bony lesions, radionuclide imaging may also play a role mostly for evaluation of extent of the disorder. We present a case of JCS evaluated with ^{99m}Tc-methylene diphosphonate (MDP) bone and ^{99m}Tc-octreotide scans.

CASE REPORT

A 27-year-old male presented to the emergency department with a fracture of the right femur following a motor accident. During imaging workup, a fracture with an underlying soft-tissue mass was detected. On histopathological examination, an NOF without any pathologic evidence of malignancy was reported. The patient underwent an orthopedic surgery for the femoral fracture. Three to four months later, following a persistent nontraumatic right arm pain, another lesion with similar pathologic result was discovered during workup. Meanwhile, the patient stated episodes of bone pain and fever as well as malaise which was gradually worsening over the preceding months. A ^{99m}Tc-octreotide scan was requested on which multiple zones with varying degrees of uptake throughout the body were noted [Figure 1]. For better localization of the lesions, a bone scan was performed subsequently which showed a similar pattern with very intense uptake in the mentioned regions [Figure 2]. Alongside, faint hyperactivity was discovered in the thorax above the region of the heart. The computed tomography scan of the chest shortly afterward

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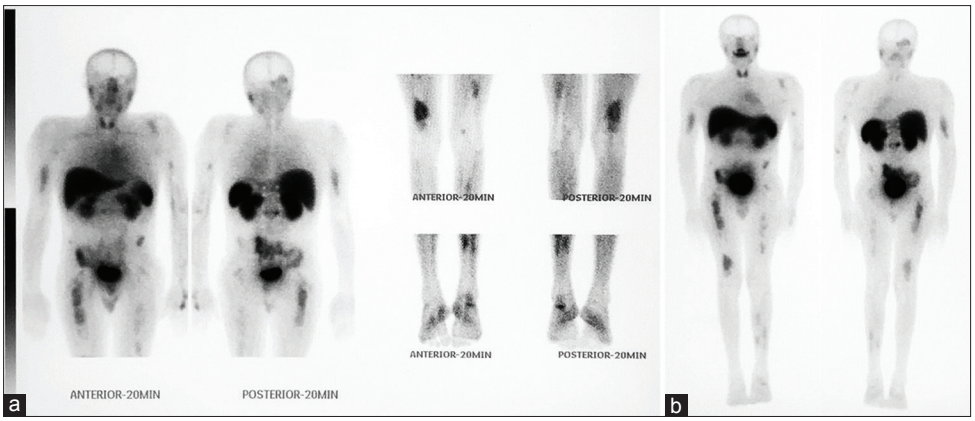

Figure 1: (a) Early phase (30 min postinjection) and (b) delayed phase (2–3 h) of whole-body ^{99m}Tc-octreotide scan of the patient. There are multiple zones with varying degrees of uptake (faint to intense) throughout the body. A region with faint uptake is evident in the mediastinum above the heart, more readily noticeable on delayed image, which is subsequently confirmed as sarcoma

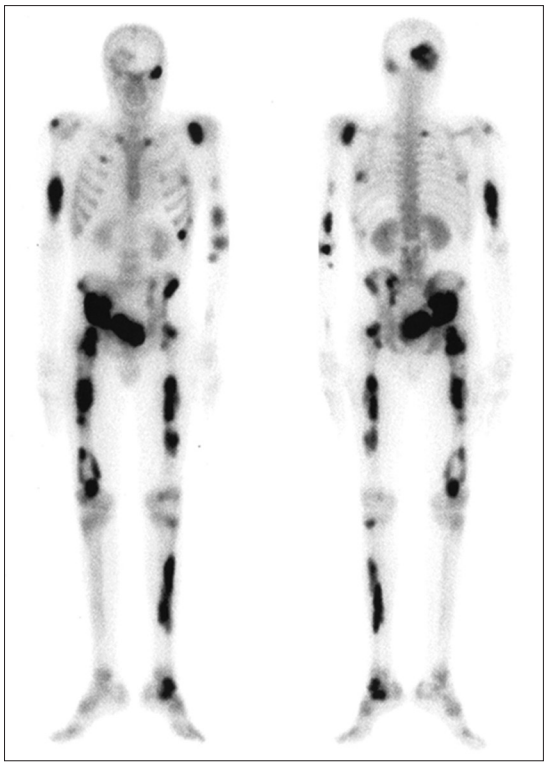

Figure 2: A whole-body ^{99m}Tc-methylene-diphosphonate bone scan is performed for better evaluation and localization of lesions. Lesions revealed similar pattern of uptake but mostly intense. The true extent of skeletal involvement on bone scan, compare to octreotide scan, is much more evident

demonstrated a mediastinal mass invading to major vascular structures. On pathology, a low-grade soft-tissue sarcoma was found, and then chemotherapy was commenced immediately. A few months later, the patient expired.

DISCUSSION

JCS is a rare genetic disorder whose etiology is not clearly understood. Historically, this syndrome was first reported by Jaffe[4] in 1958 as an entity with some different features from Albright's disease including limited skeletal fibrodysplasia, less striking skin changes (as pigmentation), and absence of sexual precocity. Later, more cases were reported with similar features by Campanacci *et al*.,[5] and Mirra *et al*.[6] In 1983, Campanacci *et al*. published a report of 10 cases with multiple NOFs and other extraskeletal anomalies in that a new syndrome with a possible connection with neurofibromatosis was proposed.[5]

Although currently, some advocate the notion of JCS as a distinct clinical entity, close pathophysiologic relationships are present with other clinical syndromes such as neurofibromatosis type 1, McCune-Albright syndrome, and Jaffe-Lichtenstein syndrome. These syndromes share common clinical and pathologic features including skeletal dysplasia and cutaneous pigmentation with JCS. However, no global consensus has been developed yet.[1,2,7,8] Since a pathogenic germline NF1 mutation has been found in a recent study in such patients, it is suggested that many JCS patients actually have neurofibromatosis.[9] Recently, a role for melatonin has also been proposed for the above spectrum of syndromes.[8] Due to this overlap, misdiagnosis, especially at presentation, is not uncommon or surprising.

JCS is clinically characterized by multiple skeletal lesions of NOF, café-au-lait spots on the skin, cardiovascular malformations, etc. Radiologically, findings compatible with NOF, although require distinguishing from polyostotic fibrous dysplasia, are evident involving the skeleton in various regions. Therefore, X-ray bone imaging seems to be the primary modality for evaluation of skeletal lesions.[1-3,10] Radionuclide imaging modalities including ^{99m}Tc-MDP bone and ^{99m}Tc-octreotide scans can be considered as adjunct tools mainly for evaluation of the true extent of the disease and possibly for biological activity of lesions. In addition, based on the degree of uptake on ^{99m}Tc-octreotide scan, a role for radionuclide peptide therapy can be considered for locally

aggressive somatostatin receptor-positive lesions to prevent pathologic fracture in weight-bearing bones. The bone scan can localize the lesion with much better clarity as in our patient. To the best of our best knowledge, no radionuclide study has been performed in the more than thirty patients with JCS reported in the literature. The multiple NOF lesions caused a debilitating morbidity to the patient, but it seems that the leading cause of death was the sarcoma in the thorax. As many reports in the literature, a pathologic fracture is a common clinical picture at presentation. To date, to the best of our knowledge, no death or concomitant malignancy is reported shortly following the diagnosis which requires a particular attention.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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