Bone Scan and SPECT/CT Findings in Marble Bone Disease

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Marble bone disease or osteopetrosis, is a rare inborn disorder characterized by the failure of osteoclasts to resorb bone [1]. Overall incidence of the disease is estimated to be 1 case in 100,000–500,000 population [2]. Whereas the radiographic features of the disease are well known, information on bone scan imaging is sparse in the literature. We present technetium-99m methylene diphosphonate (^{99m}Tc MDP) bone scan features of osteopetrosis, along with single photon emission computed tomography–computed tomography (SPECT/CT) correlation in a young male (Fig. 1).

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Fig. 1 A 23-year-old man underwent a ^{99m}Tc MDP bone scan for the evaluation of sclerotic lesions in the pelvis found on a radiograph obtained for his hip pain. On planar scan (**a**, **b**), intense tracer uptake was noted in the entire skeleton, notably in the proximal and distal shafts of bilateral femora, pelvic bones, lumbar vertebrae and proximal shafts of bilateral humeri. Considering the patient's symptoms, SPECT/CT images were obtained for radiological correlation. Coronal images of (**c**) SPECT, (**d**) CT and (**e**) fused SPECT/CT demonstrated extensive sclerosis in areas of intense tracer uptake. Most of these tracer-avid sclerotic areas were in metaphyseal regions of long bones. There was no evidence of cortical break. These findings were compatible with the diagnosis of marble bone disease or osteopetrosis. Whereas the radiographic features of OS are well known, information on bone scan imaging is sparse in the literature. On conventional imaging, generalized sclerosis—more commonly in metaphyseal regions—is seen in bones. Various patterns described on bone scan include diffuse increased uptake that may also be a “Superscan” and non-uniform uptake with foci of hyperemia [3–5]. Use of SPECT/CT in OS has not been described in literature

previously. In our case, SPECT/CT was performed to rule out fracture of left femur or pelvic bone as a cause of patient's symptoms. It is known that pelvic X-ray has limited value for detecting pelvic fractures compared with CT scanning and hence we performed SPECT/CT even when the radiographs were available [6]. In such a case, planar imaging alone will miss the uptake secondary to fracture as it will be masked by metaphyseal areas of intense tracer uptake. Additional advantage of SPECT/CT in such cases is that it can confirm that the areas of abnormal tracer uptake are limited to metaphyseal regions and hence can clinch the diagnosis of OS. In general, radiographic findings are considered sufficient for diagnosis of OS and biopsy is reserved only in pre-bone marrow transplant work-up, in cases where diagnosis is unclear or for genetic diagnosis [7]. SPECT or SPECT/CT can be used as a problem solving tool when conventional imaging findings are inconclusive or pathological fracture is suspected. Our case illustrates bone scan and SPECT/CT findings of this rare disorder. Though rare, this diagnosis should be kept in mind when similar uptake pattern and CT characteristics are encountered in practice

Conflict of Interest The authors declare no conflict of interest.

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