Scintigraphic Findings In Progressive Diaphyseal Dysplasia

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A 14-yr-old white male with a severe form of progressive diaphyseal dysplasia (Engelmann-Camurati disease) was referred to our institution for evaluation of splenomegaly, which is not usually associated with the disease. Our studies included bone, bone-marrow, and liver-spleen scans. These scintigraphic findings, along with the probable cause for splenomegaly, are discussed.

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Progressive diaphyseal dysplasia (Engelmann-Camurati disease) is a rare autosomal dominant, generalized disease of bone characterized by progressive cortical thickening with increased diameter and narrowed medullary cavities in the metaphyseal and diaphyseal regions of long bones. Epiphyses are usually spared any disease involvement and the degree of cortical thickening varies in severity. With progression the disease process may involve the calvarium, pelvis, and vertebrae.

The disease was first reported in 1922 by Camurati (*1*), but it remained for Engelmann (*2*) in 1929 to describe the specific osseous sclerosis associated with the disease. Most recently Hundley and associates (*3*) have reviewed the literature presenting 70 cases of progressive diaphyseal dysplasia, seven of which involved members of three generations of one family. Their review thoroughly covers the clinical, genetic, histologic, and radiographic findings in progressive diaphyseal dysplasia.

CASE REPORT

A 14-yr-old white male with a severe form of progressive diaphyseal dysplasia was referred to our institution for evaluation of splenomegaly discovered during a routine physical examination. Radiographically there was diffuse sclerosis of the osseous skeleton, including all the long bones of the upper and lower extremities (Fig. 1), the posterior pelvis, the lower thoracic and all lumbar vertebrae, and several skull bones. Clinically the patient exhibited the atrophy of the gluteal and proximal thigh muscles with the waddling gait and easy fatigability associated with progressive diaphyseal dysplasia.

During the evaluation, several scintigrams were made with a large-field-of-view scintillation camera, including a bone scan with 14.5 mCi of Tc-99m methylene diphosphonate (MDP), a liver-spleen scan with 5.4 mCi of Tc-99m sulfur colloid (Tc SC), and a bone-marrow scan with 10.5 mCi of Tc SC.

The findings on bone scan included markedly increased osteoblastic activity within the cortical portions of the diaphyses of all long bones, with sparing of the epiphyses, and involvement of the lower thoracic and lumbar vertebrae, pelvis, and base and right side of the skull (Fig. 2). These abnormalities matched the radiographic sclerotic abnormalities. Of note were the marrow cavities of the long bones, which seemed almost totally obliterated by the cortical thickening. This finding was complemented by a liver-spleen scan that showed a spleen measuring 15 cm on anterior and lateral images (Fig. 3) and by a bone-marrow scan that showed normal marrow uptake only in the spared epiphyses of the long bones and less than normal uptake in the pelvis, proximal halves of both femora, and the left midtibial shaft (Fig. 4). In addition, there was increased uptake within the enlarged spleen and throughout the lungs.

DISCUSSION

It has become accepted that skeletal uptake of the phosphonate complexes, including MDP, parallels both the amount of local

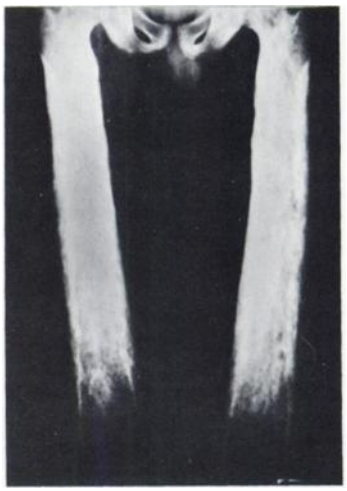

FIG. 1. Radiograph of femora showing bilateral metaphyseal and diaphyseal cortical thickening. Distal femoral epiphyses appear to be spared by the disease.

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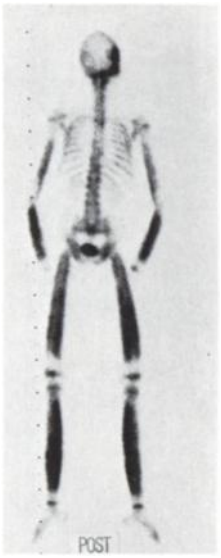

FIG. 2. Posterior whole-body bone scintigram showing involvement of pelvis, vertebrae, and ribs in addition to skull and long bones.

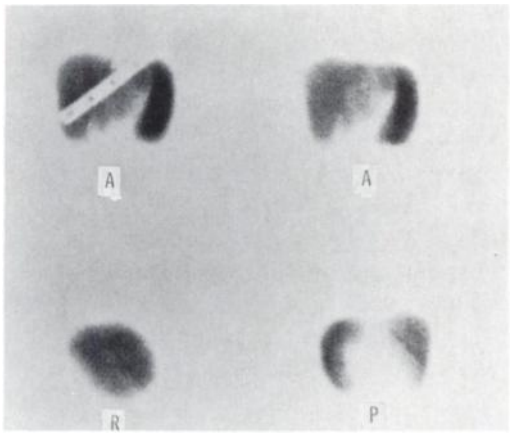

FIG. 3. Liver-spleen scintigram showing splenomegaly and increased splenic uptake of tracer. Spleen measures ~15 cm on anterior image.

blood flow and the degree of osteoblastic activity present in the bone at scanning time (*4*, *5*). In their review Hundley and associates (*3*) cite a number of cases that were studied histologically (*6–8*). One of the cardinal features appeared to be "abundant or excessive osteoblasts." These findings would explain the marked cortical thickening seen on radiograph and the markedly increased uptake of the tracer in the bone scans.

When closely scrutinizing the scintigrams of the vertebral column and the femora (Fig. 2), one can see only a narrowed trace of marrow cavity remaining in the femora. Likewise, the marrow compartment in the vertebrae is probably compromised. The bone-marrow findings parallel the bone-scan findings. In a youth of this age one should see tracer uptake in the entire femoral shafts, the proximal tibias, and the humeral shafts, in addition to the pelvis and vertebrae (*9*). This is obviously not the case in our patient. The only identifiable osseous marrow activity is present in the epiphyses of long bones, the pelvis, and focally in the proximal femora and the midshaft of the left tibia. Because of the intense tracer uptake within the enlarged spleen and the lungs, it was impossible to evaluate the marrow activity of the vertebral column; however, in

FIG. 4. Whole-body Tc-99m sulfur colloid bone-marrow scintigram showing diminished bone-marrow activity in shafts of long bones and pelvis. Only epiphyses of long bones demonstrate normal tracer uptake.

view of the sclerosis of the vertebral bodies on radiographic examination and the increased tracer uptake on the bone scan, we can assume that there is marked compromise of the bone-marrow compartment.

The patient was admitted for evaluation of splenomegaly, which is not generally seen in progressive diaphyseal dysplasia. After a thorough diagnostic evaluation it was determined that the patient's splenomegaly was due to extramedullary hematopoiesis secondary to loss of bone-marrow compartment space.

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