IMAGING CORNER

Stereotypical depiction of Erdheim-Chester Disease on Tc-99m MDP skeletal scintigraphy

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Abstract

Erdheim-Chester Disease (ECD) is a rare systemic histiocytic disorder characterized by symmetrical skeletal involvement and variable visceral manifestations. Tc-99m MDP bone scintigraphy, CT, MRI, and FDG PET-CT are essential for diagnosis and disease mapping. ECD's course depends on extraosseous involvement, with potential complications including heart and renal failure. We present the case of ECD with bilateral leg pain and orbital involvement. Imaging revealed mixed sclerotic and lytic skeletal lesions, cerebellar lesions, intraorbital soft tissue masses, mediastinal and retroperitoneal infiltration.

Keywords: Erdheim-Chester Disease (ECD), Skeletal and Visceral Involvement, Tc-99m MDP Bone Scintigraphy

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Discussion

A 55-year-old male presented to the nuclear medicine department with bilateral leg pain and prominent eyes. A Technetium-99m Methylene Diphosphonate (Tc-99m MDP) bone scintigraphy was performed, along with SPECT-CT of the lower limbs, revealing increased radiotracer uptake in the right proximal femur, bilateral distal femur, tibia, and fibula. Limited SPECT-CT correlation identified mixed sclerotic and lytic lesions without periosteal reaction. Additionally, subtle focal activity was observed along the outer margin of the left orbit and radiotracer retention was also noted in the bilateral renal collecting systems. CT imaging of the same patient demonstrated cerebellar lesions, bilateral intraconal soft tissue masses infiltrating extraocular muscles, mediastinal soft tissue encasing vascular structures, diffuse retroperitoneal soft tissue infiltration

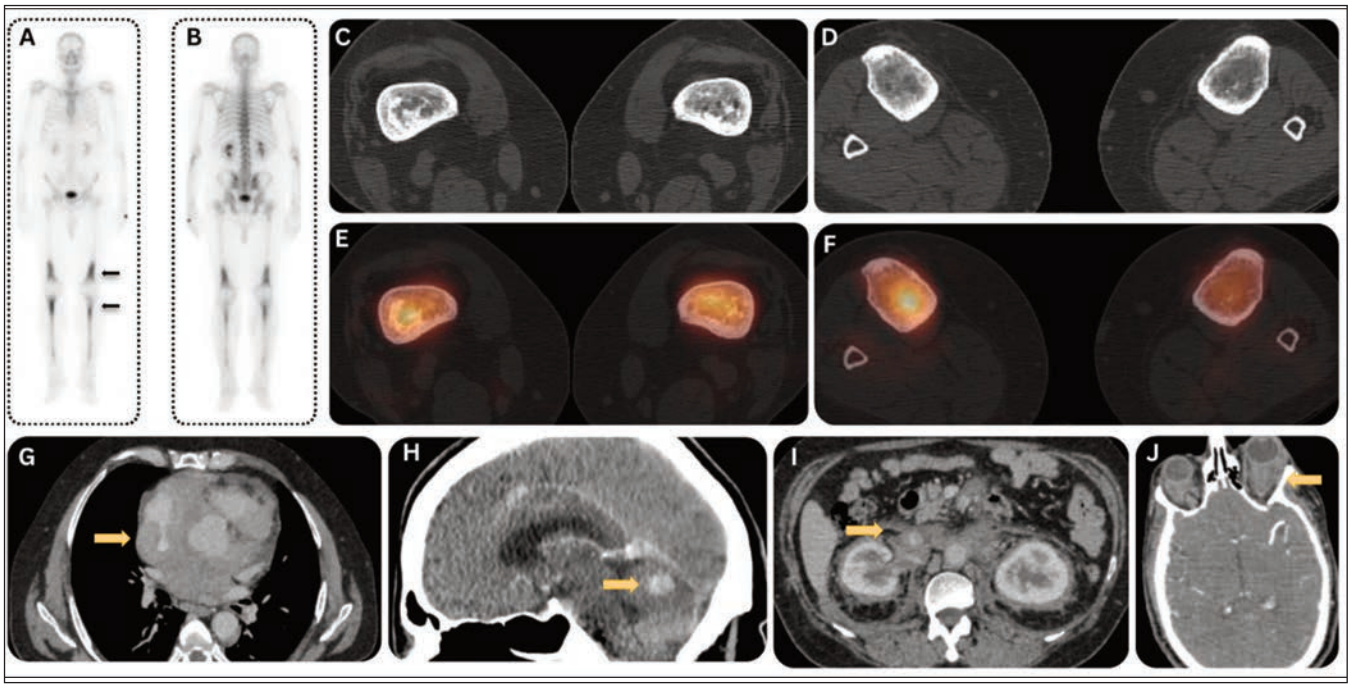

Figure: A, B) Planar whole-body scan demonstrating increased uptake in the bilateral femur and tibia (Black arrow). C, D, E, F) Mixed lytic sclerotic changes in the femur and tibia with increased uptake on the Fused SPECT-CT images. G) Mediastinal soft tissue mass encasing the vasculature (Yellow arrow). H) Cerebellar lesions (Yellow arrow). I) Retroperitoneal fibrosis invading perirenal fascia (Yellow arrow). J) Intraorbital masses more prominent on the left side (Yellow arrow).

penetrating bilateral perirenal fascia. A CT-guided core biopsy of the periaortic mass revealed inflamed fibroconnective tissue with foamy histiocyte aggregates. PCR testing for BRAF mutations was negative. These findings are diagnostic of Erdheim-Chester Disease (ECD), a rare systemic histiocytic disorder most frequently affecting males aged 40–60 years. ECD commonly involves symmetrical skeletal abnormalities in the diaphysis and metaphysis of long bones.[1,2] Tc-99m MDP bone scintigraphy provides accurate skeletal disease mapping. Visceral involvement is best assessed with computed tomography (CT), magnetic resonance imaging (MRI), and fluorodeoxyglucose (FDG) PET-CT.[3] A growing number of genetic mutations have been implicated in the pathogenesis of Erdheim-Chester Disease (ECD), notably MAPK and BRAF V600E mutation are the most frequently observed.[4] The clinical course of Erdheim-Chester Disease varies based on the extent of extraosseous and visceral involvement.

References

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