Erdheim-Chester Disease

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Erdheim-Chester disease (ECD) is a rare disease that includes sclerosis in the upper and lower extremities and extraskeletal involvement. No standard therapy exists, but interferon-α has helped some patients. The prognosis for patients with this condition is poor.

ECD is characterized by a symmetrical sclerosis at the diametaphyseal portions of the upper and lower extremities with additional extraskeletal involvement[1] including the kidney and retroperitoneum, lung, pericardium, skin, orbit, and brain. Infiltration of the pituitary stalk may lead to diabetes insipidus[2]. Skeletal involvement is characteristically bilateral and symmetric, with a characteristic bone scan finding of increased uptake in metaphyses and diaphyses of the long bones, usually sparing epiphyses.

A 56-year-old man with a history of central diabetes insipidus presented with an 8-month history of right-side leg pain. On clinical examination, exophthalmos was noted. The plain radiographs of his extremities (Figure 1) showed coarsened trabecular pattern and sclerosis mainly in the metaphyses. Delayed images in a whole-body bone scan (Figure 2) showed intense increased uptake in a bilateral symmetric distribution involving the metadiaphyseal regions of the tibias and femurs. Diagnosis of ECD was based on the results of plain radiographs and a whole-body bone scan.

Our patient was also found to have pulmonary cystic disease, 1-cm soft tissue surrounding both kidneys and the mesenteric vessels, a splenomegaly (Figure 3), and bilateral symmetrical enhancing soft tissue mass within the superior extraconal aspect of both orbits (Figure 4). The diagnosis of ECD was confirmed histologically by a kidney biopsy. It showed dendritic cells rich in a perirenal fibrotic process with lipid-laden histiocytes and Touton-type giant cells, which were positive for CD68 and negative for S-100, consistent with a diagnosis of ECD.

The prognosis of patients with ECD is poor. The survival of patients at 3 years is about 59%. The causes of mortality

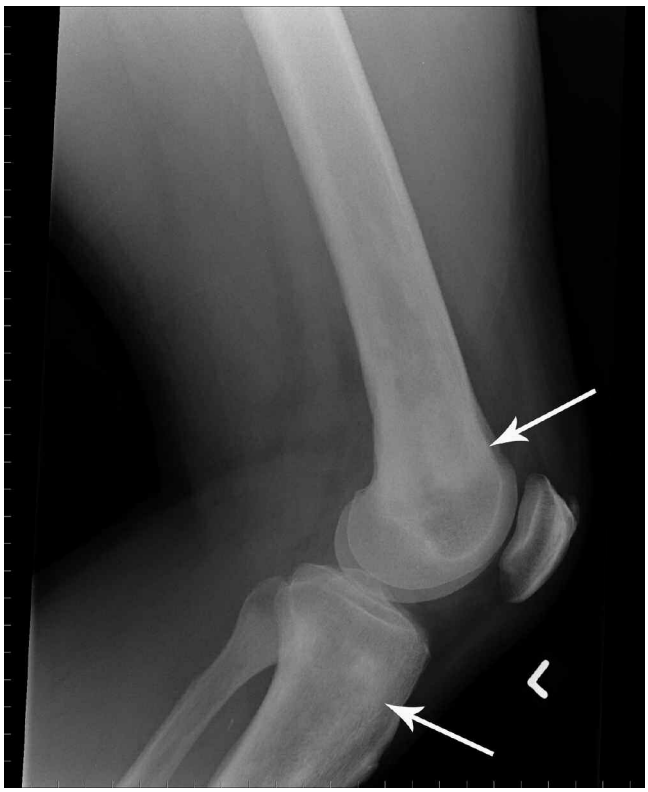

Figure 1. Within the distal femur and proximal tibia is a diffuse symmetric diametaphyseal process showing mixed sclerosis (arrows) and slight lucency.

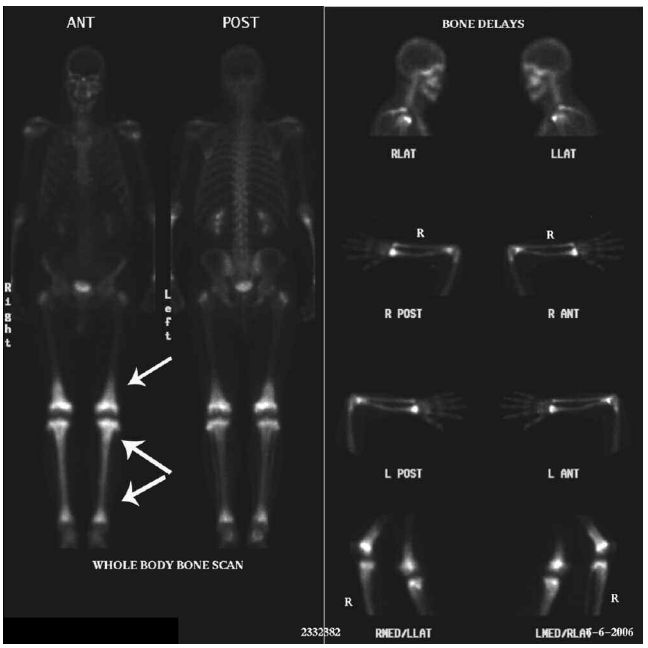

Figure 2. Delayed image in a whole-body bone scan shows intense increased uptake in a bilateral symmetric distribution involving the metadiaphyseal regions of the tibias and femur (arrows). To a lesser extent, similar findings are noted in the forearms.

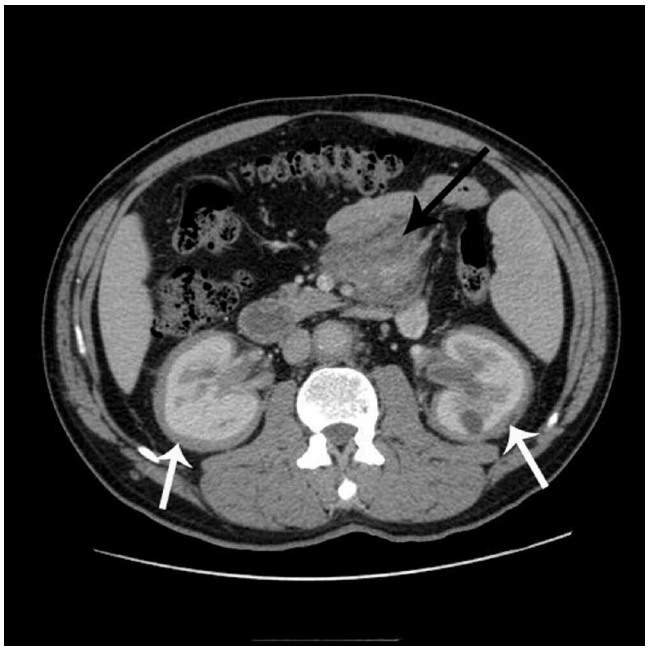

Figure 3. Axial image of a contrast-enhanced computed tomography abdomen scan shows 1-cm soft tissue surrounding both kidneys (white arrows). A soft tissue mass surrounds the mesenteric blood vessels (black arrow).

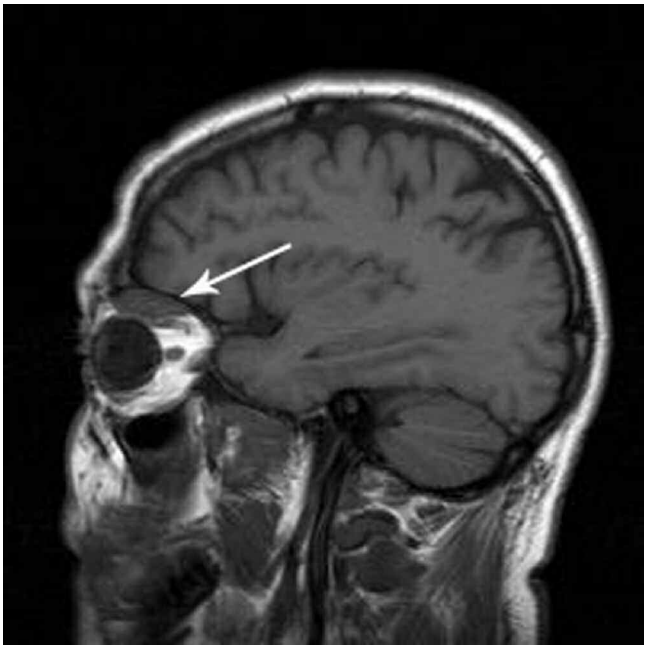

Figure 4. Sagittal T1-weighted image of the brain shows a soft tissue mass in the superior extraconal aspect of the orbit (arrow). This is seen bilaterally.

are often from respiratory distress, cardiac failure, or pulmonary fibrosis[2].

There is no standard therapy for this disorder, although responses to interferon-α have been reported in some patients. Bisphosphonates and steroid have shown some positive effect for bony pain[2]. Our patient has now been treated with interferon-α 2b for 1.5 years. On this treatment, his leg pain has mostly disappeared, with improving exophthalmos.

REFERENCES

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