MAYO CLINIC

Erdheim-Chester Disease

Mithun Vinod Shah, MD, PhD, and Ronald S. Go, MD

From the Division of Hematology, Mayo Clinic, Rochester, MN.

Scan code to view additional images

A 57-year-old man presented with flank pain, temporal headaches, and leg claudication. He was found to have new-onset hypertension and acute kidney injury. Subsequent evaluation revealed bilateral hydronephrosis due to an intense bilateral perirenal and retroperitoneal fibrotic process, giving rise to "hairy kidney" appearance (Figure 1, white arrow). Fibrotic involvement was also seen throughout the length of the major arteries including the aorta (Figure 1, "coated aorta," black arrow) and the femoral arteries (Supplemental Figure, panel A, available online at http://www.mayoclinicproceedings.org), leading to femoral arterial occlusions. Bone scan highlighted bilaterally symmetrical intense tracer uptake in the metaphyses and epiphyses of the long bones, which was more prominent in the lower limbs (Figure 2). Given characteristic findings, the diagnosis of Erdheim-Chester disease (ECD) was considered.

Erdheim-Chester disease is a rare multisystem histiocytic disorder that commonly affects the bones, the central nervous system (diabetes insipidus, ataxia), heart and great vessels, kidneys, skin (xanthoma), and retroperitoneum. Microscopic features of ECD include dense fibroadipose tissue with foamy non-Langerhans' cell histiocytes (Touton-like giant cells), diffusely infiltrating the surrounding tissue (Supplemental Figure, Panel B, $CD68^{+}/CD163^{+}/S100^{-}/CD1a^{-}$, available online at http://www.mayoclinicproceedings.org).

Generally, immunomodulators such as corticosteroids, methotrexate, interferon, or chemotherapeutic agents such as cladribine (2-CdA) have been used with limited success. Polymerase chain reaction assay from the perinephric tissue biopsy showed *BRAF* V600E mutation, compatible with the diagnosis of ECD. Given the presence of *BRAF* V600E, the patient is being treated with a clinical trial of vemurafenib, a BRAF inhibitor.

Correspondence: Ronald S. Go, MD, Division of Hematology, Mayo Clinic, 200 First St SW, Rochester, MN 55905 (Go.Ronald@mayo.edu).

SUPPLEMENTAL ONLINE MATERIAL

Supplemental material can be found online at http://www.mayoclinicproceedings.org. Supplemental material attached to journal articles has not been edited, and the authors take responsibility for the accuracy of all data.

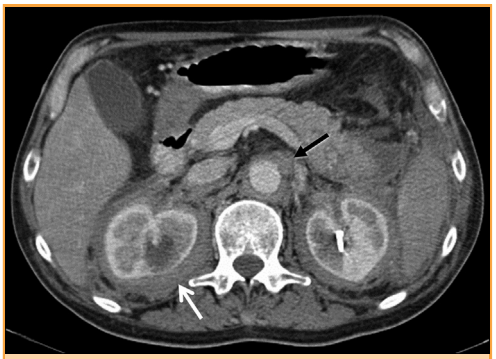

FIGURE 1. "Hairy kidneys" and "coated aorta" characteristic of Erdheim-Chester disease.

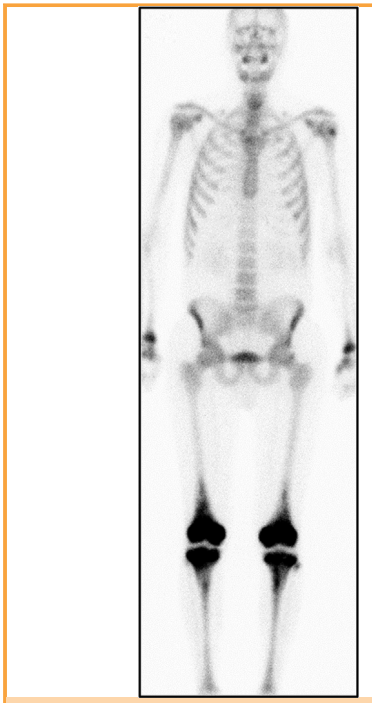

FIGURE 2. Bilateral symmetric osteosclerosis of the long bones of lower limbs.

Mayo Clin Proc. ■ September 2015;90(9):1310 ■ http://dx.doi.org/10.1016/j.mayocp.2015.03.019
www.mayoclinicproceedings.org ■