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Erdheim-Chester Disease: Computed Tomography in Two Cases

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Erdheim-Chester disease is an unusual lipoidosis in which characteristic radiographic features have been described in the skeleton [1-3]. Extraosseous manifestations have been reported in the retroperitoneum, lungs, and heart. We report two cases of Erdheim-Chester disease with orbital involvement demonstrated by computed tomography (CT).

Case Reports

Case 1

A 70-year-old man presented with painless, progressive protrusion of both eyes of 3 months' duration. He had been hospitalized on several occasions for dyspnea, congestive heart failure, and weight loss. Bronchial and pleural biopsy demonstrated fat-laden macrophages, but a definite diagnosis was not established. CT of the orbits demonstrated bilateral enhancing retrobulbar masses (fig. 1A). The mass in the right orbit diffusely infiltrated the retroorbital fat and obscured the normal architecture of the orbit. The optic nerve could not be discretely identified. On the left side a more focal mass was located mainly lateral to the optic nerve and did not reach the orbital apex. Bilateral exophthalmos was present. The extraocular muscles

appeared normal in size. The rest of the CT scan of the head was normal. A right orbitotomy was performed and a biopsy obtained. The pathologic interpretation of the tissue by the Armed Forces Institute of Pathology (AFIP) was that of an unusual type of xantho-granulomatous inflammatory pseudotumor. A trial course of prednisone had no effect on the retrobulbar masses.

Two years later, the patient was hospitalized with congestive heart failure. Bone radiographs demonstrated symmetric areas of sclerosis in the metaphyses of the long tubular bones (fig. 1B). He was treated for cardiac decompensation and recovered. His eyes remained stable. Fourteen months later, at age 74, he developed right-sided heart failure secondary to pulmonary fibrosis and died. A review of tissue obtained from the head of the right femur revealed focal infiltration by large, clear, xanthomatous histiocytes and discrete collections of cholesterol clefts surrounded by multinucleated foreign-body giant cells. The combination of radiographic and histologic findings was believed to represent Erdheim-Chester disease.

Case 2

A 48-year-old woman was diagnosed by the AFIP as having Erdheim-Chester disease on the basis of long-bone radiographs and



Fig. 1.—Case 1. A, Contrast-enhanced orbital CT scan. Bilateral enhancing retroorbital masses. Diffuse infiltration of retroorbital fat on right side; more focal infiltration on left (arrows). B, Long-bone radiograph 2 years later. Sclerotic areas in femur and tibia.

Fig. 2.—Case 2. Contrast-enhanced orbital CT scan. Diffuse infiltration of retroorbital fat bilaterally, causing exophthalmos.

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bone biopsy. Retroperitoneal fibrosis was diagnosed by excretory urography and presumed to be secondary to Erdheim-Chester disease. She also had bilateral exophthalmos and papilledema. CT of the orbits with intravenous contrast medium demonstrated diffuse soft-tissue infiltration of the retrobulbar areas of both orbits (fig. 2). The abnormal tissue was of higher attenuation than brain parenchyma, probably reflecting contrast enhancement. The extraocular muscles and optic nerves could not be discretely visualized. Marked exophthalmos was present. The rest of the head scan, including the sella and parasellar areas, was normal. The patient died 18 months later, and the autopsy report of massive lipogranulomatosis of the orbit, retroperitoneal tissues, mesentery, pericardium, and skin confirmed the diagnosis of Erdheim-Chester disease.

Discussion

The diagnosis of Erdheim-Chester disease can be made confidently when the characteristic radiographic findings are seen in combination with the pathologic demonstration of foamy, cholesterol-laden histiocytes in the bone marrow, surrounded by sclerotic bone [1, 3]. Clinical manifestations of the disease are variable and nonspecific. Highly suggestive radiographic features of the disease have been described and include bilateral, usually symmetric, sclerosis of the long tubular bones of the appendicular skeleton. Extraskelatal involvement has been reported in the retroperitoneum, lungs, pericardium, myocardium, and skin [2-4]. The typical involvement of the long tubular bones of the skeleton has been present in all reported cases of extraskelatal involvement and was present in our two cases as well.

The bilateral orbital involvement in our cases occurred as one manifestation of a severe diffuse systemic disorder. However, as case 1 illustrates, orbital involvement may be the source of the initial complaint. An important difference between case 1 and case 2 was the focal mass involving the

left orbit in case 1, demonstrating that the disease may appear as a discrete mass and that asymmetry can also occur. Involvement of the optic canals, pituitary gland, optic chiasm, and parasellar areas was specifically sought but could not be demonstrated; however, the autopsy 18 months after CT in case 2 revealed infiltration around the pituitary gland. There was no evidence of lipogranulomatous infiltration of the brain on CT or at autopsy.

The differential diagnosis of the orbital manifestation of Erdheim-Chester disease is primarily that of inflammatory pseudotumor, thyroid eye disease, lymphoma, and metastatic disease. A definite diagnosis of Erdheim-Chester disease cannot be made on the basis of the orbital CT scan alone, but the radiologist should suggest the diagnosis and recommend the appropriate radiographs. A full understanding of the nature of the infiltrative process and the variety of clinical and radiologic features together with a high index of suspicion will enable the radiologist to interpret a confusing clinical presentation.

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