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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 xanthogranulomatous 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...
bone biopsy. Retroperitoneal fibrosis was diagnosed by excretory
urography and presumed to be secondary to Erdheim-Chester dis-
ease. 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 paren-
chyma, 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 con-
firmed 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, sur-
rrounded 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. Extraskeletal
involvement has been reported in the retroperitoneum, lungs,
pericardium, myocardium, and skin [2–4]. The typical involve-
ment of the long tubular bones of the skeleton has been
present in all reported cases of extraskeletal 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. How-
ever, as case 1 illustrates, orbital involvement may be the
source of the initial complaint. An important difference be-
tween 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 recom-

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