LETTER / neuroradiology

Erdheim-Chester disease: A rare diagnosis with evocative imaging

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Observation

A 60-year-old male patient was examined for the exploration of bilateral retro-orbital tumours. These tumours were discovered two years earlier in a brain scan for the exploration of an exophthalmous. Two biopsies of these lesions were taken. The first one revealed a polymorphous inflammatory tissue. The second one revealed a strong granulomatous reaction, rich in non-specific foamy histiocytes. During his hospitalisation, the patient described a weight loss of 12 kg over the last year as well as diffuse periodic inflammatory bone pain. The thoracic-abdominal-pelvic CT scan detected peri-aortic infiltration with the typical appearance of an aortic sleeve (Fig. 1) associated with "hairy" kidneys (Fig. 2). The association of these signs pointed to a diagnosis of Erdheim-Chester disease, confirmed by a bone scan (Fig. 3) and the re-examination of the histological samples.

Discussion

First described by Chester in 1930 [1], Erdheim-Chester disease is a rare non-Langerhans histiocytosis with a specific tropism for connective fatty and perivascular tissue. The cause is not known. The diagnosis of this systemic disease is histological, revealing a tissue infiltration by foamy histiocytes, without cytoplasmic Birbeck granules, associated with...
Figure 1. Thoracic scan with injection of contrast product in mediastinal opening. Homogenous and circumferential peri-aortic infiltration (aortic sleeve) (arrows).

Figure 2. Abdominal-pelvic scan with injection of contrast product. Infiltration of the “hairy” peri-renal fat (arrows).

Figure 3. Bone scintigraphy: a: static tissue time image centred on the legs revealing hyperhaemia of the right and left tibia; b: static bone image centred on the legs individuating heterogeneous fixation of the right and left tibia.
a specific immuno-histochemical profile with positive CD68 labelling while the CD1a and Protein S100 markers are negative, as opposed to histiocytosis X [2, 3]. Men are more likely to have the disease, with a peak occurrence between 40 and 60 years of age. The clinical polymorphism makes the diagnosis difficult.

General signs are often found (fever, asthenia, weight loss). Diffuse or localised bone pain is found in most cases. Orbit impairment such as exophthalmia is a classic means of detection. Pulmonary (cough, dyspnoea), gastric (pain), neurological (pyramidal syndrome, cerebellar syndrome, etc.) and endocrine signs (insipid diabetes by infiltration of the hypophyseal stalk) are often associated although none of them are specific. In the patient, the CT was carried out for retro-orbital xantho-granulomatous tumours. Retroperitoneal impairment, the most common extra-osseous location in Erdheim-Chester disease was present in the form of bilateral and symmetrical tissue infiltration of the peri-renal fat producing an appearance of “hairy” kidneys, suggestive of the disease. This impairment may evolve in the form of retroperitoneal fibrosis, with coating of the kidney sinus and dilation of the pelvicalyceal system [4]. Of course, the differential diagnosis is renal lymphoma, certain forms of which correspond to a homogenous, possibly bilateral, peri-renal mass. However, images of peri-renal stranding, when important, may raise diagnostic problems.

If in doubt, a biopsy may be taken [5]. The other typical impairment is the appearance of aortic sleeve, resulting in a circumferential and homogenous peri-aortic infiltration. This is a rare and poorly known location, although evocative [6]. Bone impairment is constant in this disease. There are two fully distinct forms: the so-called “classic” form, associating condensation of the heterogeneous spongy bone and the filling of the medullary cavity of long bones. The second, more atypical, impairment is the pseudo-tumoral lytic appearance involving a difficult diagnosis. Our patient suffered from diffuse bone pain without visible radiographic lesion. However, the bone CT scan found a bilateral and symmetrical tibial hyperfixation, fully evocative of Erdheim-Chester disease. Moreover, it differs from other histiocytoses, that do not fix technetium 99m in scintigraphy [7]. Damage to the pulmonary parenchyma is not noted, and this is rare in this disease. The most classic manifestations are micronodulation of lymphangitic distribution with cysts and sometimes pseudo-tumoral pelvicalyceal impairment such as thickening [6]. Moreover, the encephalic MRI did not detect any anomaly. Infiltration of the hypophyseal stalk may account for possible diabetes insipidus, which was not found in our patient [8].

Conclusion

Erdheim-Chester disease is a rare systemic disease. The diagnosis is difficult due to the major clinical and morphological polymorphism. The retroperitoneal impairment with the appearance of “hairy” kidneys suggests the disease, as does aortic infiltration. The diagnosis is always histological, revealing foamy histiocytes presenting a specific immuno-histochemical profile.

Disclosure of interest

The authors declare that they have no conflicts of interest concerning this article.

References