Case 13417
Erdheim-Chester disease

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Section: Musculoskeletal System
Published: 2016, Sep. 12
Patient: 73 year(s), male

Clinical History

A 73-year-old man was referred to the internal medicine department. His complaints were lifelessness and physical deconditioning since one and a half year. Laboratory results showed microcytic anaemia and raised inflammatory markers. A gastroscopy, colonoscopy and bone marrow aspiration could not reveal any suspicious findings. 18FDG-PET/CT imaging was performed.

Imaging Findings

CT imaging of the legs shows medullar osteosclerosis with cortical thickening, affecting the diaphysis and metaphysis. High FDG uptake in the corresponding areas on 18FDG-PET imaging (1a, b). CT imaging of thorax and abdomen shows soft tissue infiltration around the aorta (2a-e), expanding into the left and right paravertebral space (2a-c). Note the retrosternal (2a) and pleural soft tissue infiltration (2b). The adrenal glands are swollen with soft tissue infiltration of the adjacent fat. Increased FDG tracer uptake in the adrenal glands, around the aorta and in the stomach (2d). Soft tissue infiltration in the left and right perirenal and posterior pararenal space ('hairy kidney' sign), expanding into the renal sinuses (2e). A left-sided hypermetabolic spot anterior to the middle cerebral artery and a high FDG tracer uptake in the small bones of the feet on 18FDG-PET imaging, with no corresponding lesions on CT imaging (3a, b).
Discussion

Erdheim-Chester Disease (ECD) is a rare multisystemic disease of unknown aetiology [1, 2]. It is characterized by a typical pattern of osseous infiltration and a varying degree of extraskeletal involvement [3, 4]. Most frequent symptoms are bone pain, diabetes insipidus, exophthalmos, renovascular hypertension or neurological manifestations [4-6].

Osseous infiltration generally consists of a symmetric bilateral osteosclerosis in the metaphysis and diaphysis of the long bones [2, 3, 5]. Other radiographic features are osteolysis, cortical thickening, narrowing of the medullary cavity, a blurred corticomedullary margin, a metaphyseal radiolucent band and signs of periostitis. On MR imaging, osteosclerosis is hypointense on T1-weighted images and hyperintense on T2-weighted images; enhancing after gadolinium injection [3]. Bilateral soft tissue infiltration of the perirenal and posterior pararenal space is called the 'hairy kidney' sign. This sign consists of a homogeneous band with spiculated contours, hypattenuating on CT imaging and isointense to muscle on T1 and T2 weighted MR imaging [2, 4]. Infiltration can expand into the adrenal fossa [4, 5]. Diffuse circumferential involvement of the aorta is called the 'coated aorta', hypo-attenuating on CT imaging and isointense to muscle on T1- and T2-weighted images [2, 4]. Cardiac involvement manifests as pericardiac soft tissue thickening, pericardiac effusion or a myocardial pseudo-tumorous mass. On high resolution CT imaging smooth septal thickening, centrilobular nodules or pleural thickening can be seen [2, 4, 5]. Retro-orbital soft tissue involvement is usually intraconal, bilateral and lacks signal intensity on T1 and T2 weighted MR imaging. Hypothalamic-pituitary axis involvement appears on T1 weighted MR imaging as a nodular mass in the pituitary stalk or an absence of signal in the posterior pituitary [5]. Neurological involvement can manifest as nodules or mass lesions in the brain, the meninges or the spinal cord with an increased signal intensity on T2 weighted MR imaging and prolonged enhancement after gadolinium injection on T1 weighted MR imaging [6, 7].

99mTc bone scintigraphy and 18FDG-PET/CT imaging have the advantage of performing a whole-body examination [3, 8]. 99mTc bone scintigraphy will demonstrate an increased tracer uptake in all bony lesions and can detect radiographically silent bone involvement [3]. 18FDG-PET/CT imaging demonstrates a high FDG uptake in all metabolically active lesions [8].

Diagnosis of ECD is suspected on imaging findings and confirmed by histological analysis of a biopsy. Globally, prognosis is poor and therapeutic options are scarce. Medical treatment includes corticosteroids, bisphosphonates, cytotoxic agents and immunosuppressive drugs [2, 5]. Surgical intervention is indicated when a mass compresses the brainstem or the spinal cord [6].

Final Diagnosis

Diagnosis of Erdheim-Chester Disease was confirmed on a bone biopsy.

Differential Diagnosis List

Langerhans cell histiocytosis, Rosai-Dorfman disease, Paget's disease, Idiopathic retroperitoneal fibrosis [9]
Medullar osteosclerosis, cortical thickening and corticomedullar blurring of the left and right femur (upper figure, arrows). 18FDG-PET scanning reveals high FDG tracer uptake in the medulla of the left and right femur (lower figure, arrows).

Area of Interest: Bones; Extremities; Musculoskeletal bone;
Imaging Technique: CT; PET;
Procedure: Diagnostic procedure;
Special Focus: Connective tissue disorders;

Medullar osteosclerosis of the left femur and tibia. Corresponding high FDG tracer uptake (arrows). Normal appearance of the epiphyses (left figure, arrowheads). Increased FDG tracer uptake in calcaneus, cuboid bone and metatarsals (right figure, arrow).
Infiltration of the retrosternal soft tissues (thick arrow) and infiltration of the soft tissues around the thoracic aorta (thin arrow) with expansion into the left and right paravertebral space.

Infiltration of pleura posterior to the left ventricle (thick arrow) and of the soft tissues around...
the thoracic aorta (thin arrow) with expansion into the left and right paravertebral space.

Area of Interest: Arteries / Aorta; Respiratory system;
Imaging Technique: CT;
Procedure: Diagnostic procedure;
Special Focus: Connective tissue disorders;

Soft tissue infiltration around the thoracic aorta (arrow) with expansion into the left and right paravertebral space.

Area of Interest: Arteries / Aorta;
Imaging Technique: CT;
Procedure: Diagnostic procedure;
Special Focus: Connective tissue disorders;

Adrenal gland swelling with soft tissue infiltration of the adjacent fat and around the aorta (arrows). High FDG tracer uptake in the adrenals, around the aorta (arrows) and in the stomach (long arrow).

Area of Interest: Adrenals; Arteries / Aorta; Stomach;
Imaging Technique: CT; PET;
Procedure: Diagnostic procedure;
Special Focus: Connective tissue disorders;
Soft tissue infiltration of the left and right perirenal and posterior pararenal spaces (thin arrows). Infiltration of the left and right renal sinus (thick arrows). Infiltration around the abdominal aorta.

Figure 3 PET imaging

Axial view of the brain shows a left-sided hypermetabolic spot anterior to the middle cerebral artery.
Axial view of the feet shows a high FDG tracer uptake in the left and right calcaneus (upper figure, arrows), cuboid bone (middle figure, arrows) and metatarsals (lower figure, arrows).

References


manifestations and neuroradiological presentation of Erdheim-Chester disease: report of 6 cases and systematic review of the literature. J Neurol 253(10):1267-77


Citation

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Erdheim-Chester disease {Online}
URL: http://www.eurorad.org/case.php?id=13417