Case 14798

Eurorad

An unusual infiltrative disease of the brain: the Erdheim-Chester disease

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Section: Neuroradiology
Area of Interest: Neuroradiology brain Cardiovascular system Lung Gastrointestinal tract Bones
Procedure: Diagnostic procedure
Procedure: Defecography
Imaging Technique: CT
Imaging Technique: MR
Imaging Technique: Nuclear medicine conventional
Special Focus: Haematologic diseases Case Type: Clinical Cases
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Patient: 43 years, male

Clinical History:

A 43-year-old male patient was admitted in our hospital for palpitations, cervical pain and dyspnoea. The patient complained of several transient episodes of disorientation in the last two months. Also, the patient reported myoclonus, intrusive thoughts and the symptoms of central diabetes insipidus as polyuria and polydipsia.
Imaging Findings:

The brain CT described multiple hypodense lesions localized in the pons, which show a diffuse and inhomogeneous contrast enhancement. The orbits were occupied by hypodense, bilateral, pseudotumoral masses, with intraconal extension. The same lesions displaced the extrinsic muscle and the optic nerves. After the injection of contrast medium, the orbital masses showed a homogenous and rapid enhancement (Fig. 1).

For a better definition of brain involvement, we performed also the brain MRI, which showed multiple areas of high intensity signal in T2 sequences, localized in the hippocampus, parahippocampal gyri, pons and middle cerebellar peduncoles. The lesions infiltrated also the hypothalamus with extension to tuber cinereum. All of these lesions enhanced after the injection of gadolinium (Fig. 2). The brain MRI confirmed the presence of bilateral pseudotumoral masses in the orbits, which appeared hypointense in T1 and T2 sequences with homogeneous contrast enhancement (Fig. 3).
Discussion:

Erdheim-Chester disease (ECD) is a rare non-Langherans cell histiocytosis, first described in 1930. The disease is characterized by the infiltration of tissues by foamy non Langerhans-cell histiocytes CD68+, CD1a-, S-100-, with middle aged male predominance [1].
The most common site of involvement is the distal diaphysis of long bones, in particular the femurs [2]. However, the

disease may infiltrate the retroperitoneum, the lungs, the cardiovascular system and the central nervous system [3].

The brain infiltration occurs in about 40% of patients. The most frequent intracranial site involved is the hypothalamic-pituitary axis. The MRI is the modality of choice to identify hypothalamic and pituitary lesions. They appear hyperintense in T2 sequences and enhance after the injection of gadolinium. The hyperintensity of the posterior pituitary lobe on T1 sequences may be absent. The pituitary involvement may cause the central diabetes insipidus, frequently seen in Erdheim-Chester disease [4].
Other intracranial parenchymal lesions occur more frequently in the infratentorial region. They are hypodense at CT and hyperintense in T2 sequences at MRI, with variable enhancement and localizations. They may occur in the brainstem, cerebellum, hippocampus, mammillary bodies, fornix, cerebellar peduncles and parahippocampal gyri [5].
The supratentorial infiltration is less commonly observed [1]. The clinical manifestations are dependent on the anatomical localization of the lesions and the cerebellar ataxia and the pyramidal signs are the most frequent symptoms [4]. The meningeal lesions are sometimes observed and are often single or multiple dural meningioma-like masses or stellate formations [4-6]. The spinal cord lesions are very rare [7].
About 30-40% of patients with Erdheim-Chester disease have orbital involvement, which may cause exophthalmos. The orbital lesions are usually bilateral, intraconal xanthogranulomatosus pseudotumours, but unilateral masses are also reported. At MRI, they appear hypointense in T1 and T2 sequences with homogeneous enhancement after injection of gadolinium [8].

In our case, along the neuroradiological evaluation, we performed also a thoraco-abdominal CT study, which showed the involvement of cardiovascular system, lungs and the presence of retroperitoneal fibrosis. These features and the brain infiltration direct us towards the diagnosis of a systemic infiltrative disease, like Erdheim-Chester disease. To support this hypothesis, we performed also the bone scintigraphy, which demonstrated the infiltration of the distal right femur. However, we performed a CT guided biopsy of retroperitoneal mass and the histological proofs showed the tissue infiltration by non-Langerhans cell histiocytes. This finding confirmed the diagnosis of Erdheim-Chester disease.
The patient was treated with steroids and interferon alpha with a good clinical response.
Differential Diagnosis List: Erdheim-Chester disease, IgG4-related disease, Langerhans cell histiocytosis

Final Diagnosis: Erdheim-Chester disease

References:

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Takeuchi T, Sato M, Sonomura T, Itakura T. (1989) Erdheim-Chester disease associated with intramedullary spinal cord lesion. Br J Radiol 85(1011):e62-4 (PMID: 22391503)
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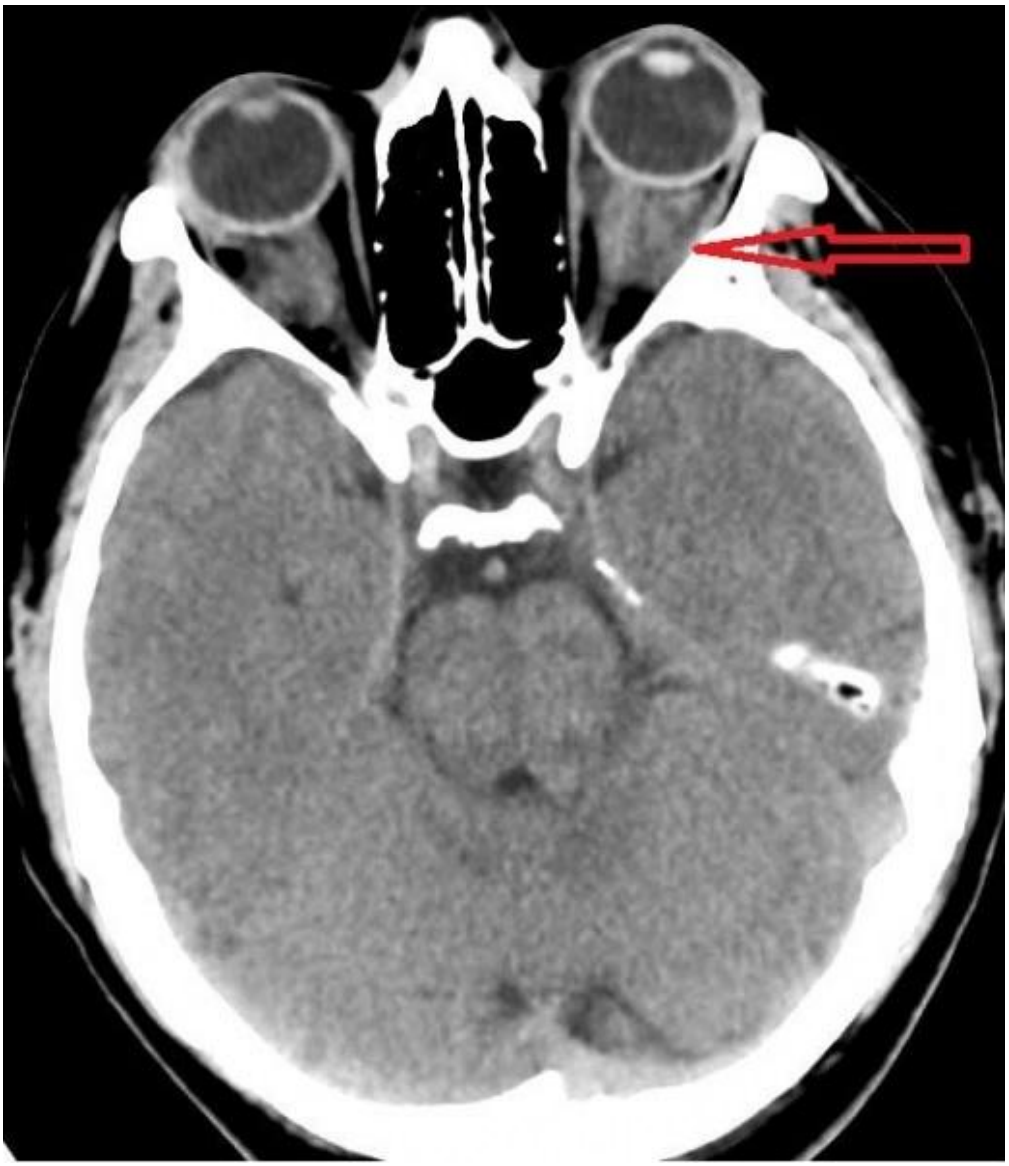
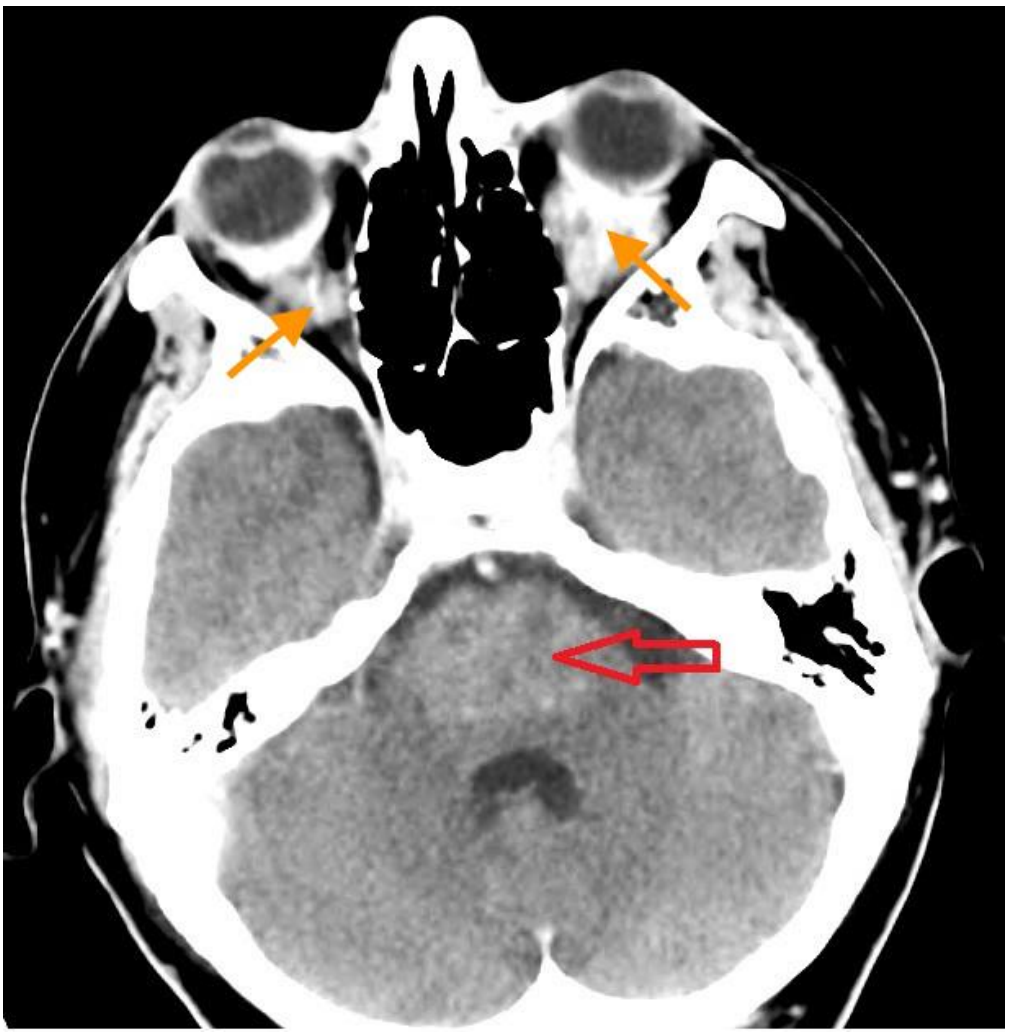
Figure 1

a



Description: Pre-contrast axial CT image shows bilateral retrobulbar pseudotumours with intraconal extension (red arrow). The optic nerves and extrinsic muscles are delocalized by the masses. **Origin:** Department of Radiology, San Paolo Hospital, Milan, Italy

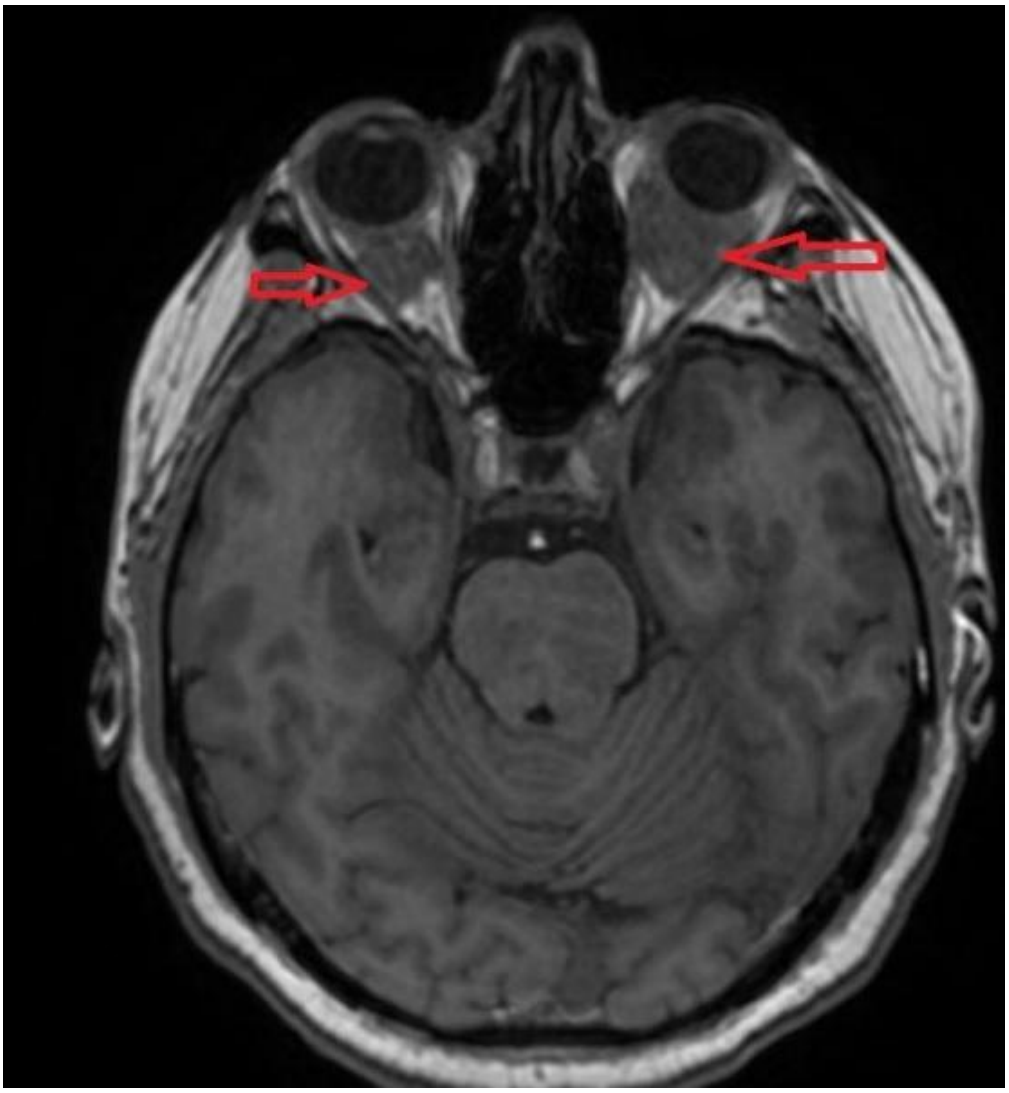
b



Description: The post-contrast images show a homogeneous enhancement of the retrobulbar masses (orange arrows). Note also the inhomogeneous enhancement in the pontine region due to histiocyte infiltration (red arrow). **Origin:** Department of Radiology, San Paolo Hospital, Milan, Italy.

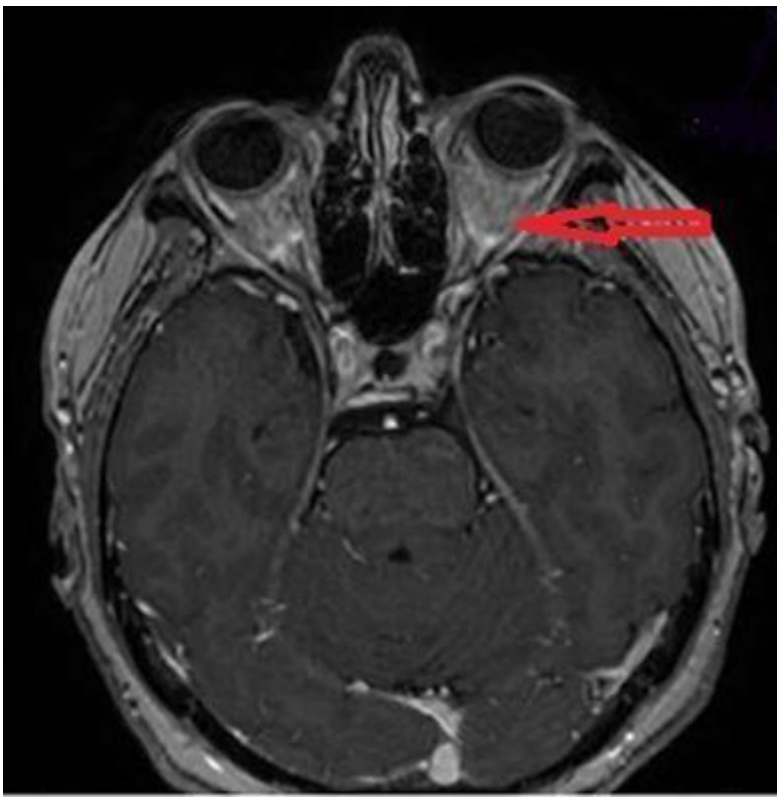
Figure 2

a



Description: The axial T1 image shows the presence of bilateral hypointense pseudotumoral masses in the retrobulbar space, which replace the normal fat (red arrows). **Origin:** Department of Radiology, San Paolo Hospital, Milan Italy

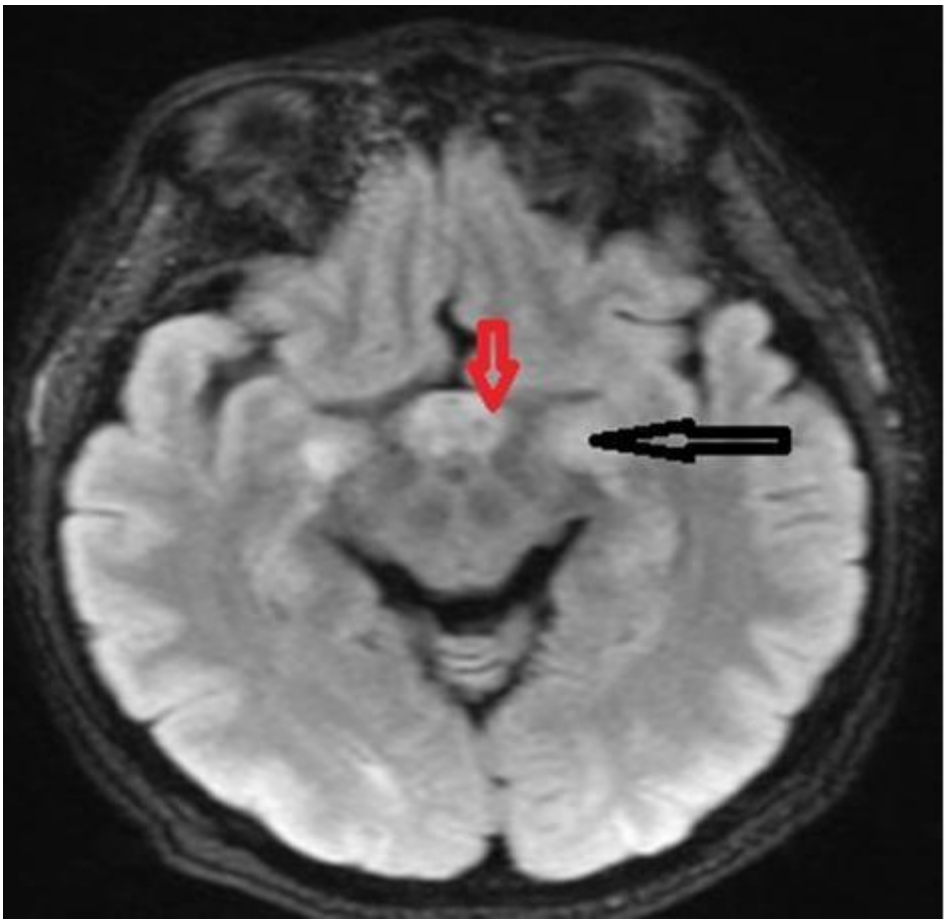
b



Description: After the admnistration of gadolinium, the masses in the retrobulbar space show a homogenous enhancement (red arrow). **Origin:** Department of Radiology, San Paolo Hospital, Milan, Italy

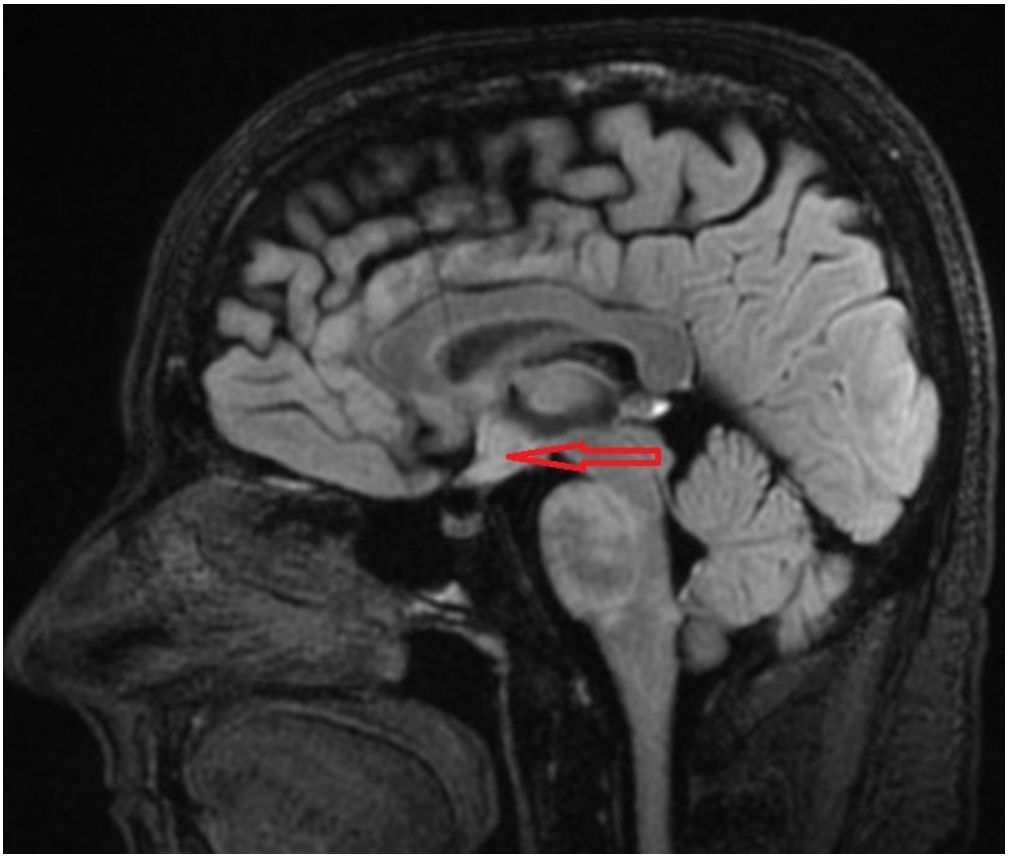
Figure 3

a



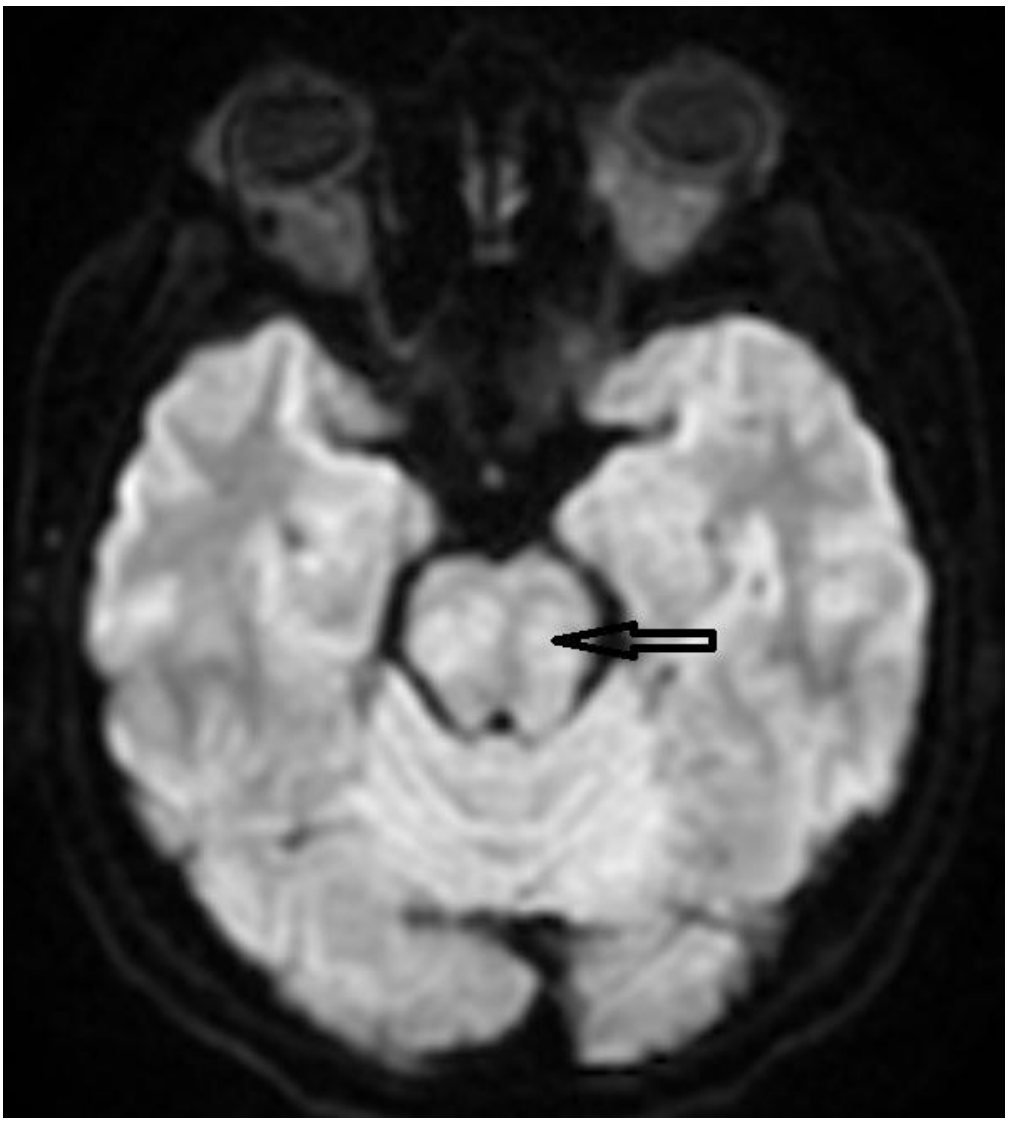
Description: The T2 FLAIR image shows the presence of hyperintense lesions in the hippocampus and parahippocampal gyri (black arrow). The disease infiltrates also the hypothalamic region (red arrow). **Origin:** Department of Radiology, San Paolo Hospital, Milan, Italy.

b



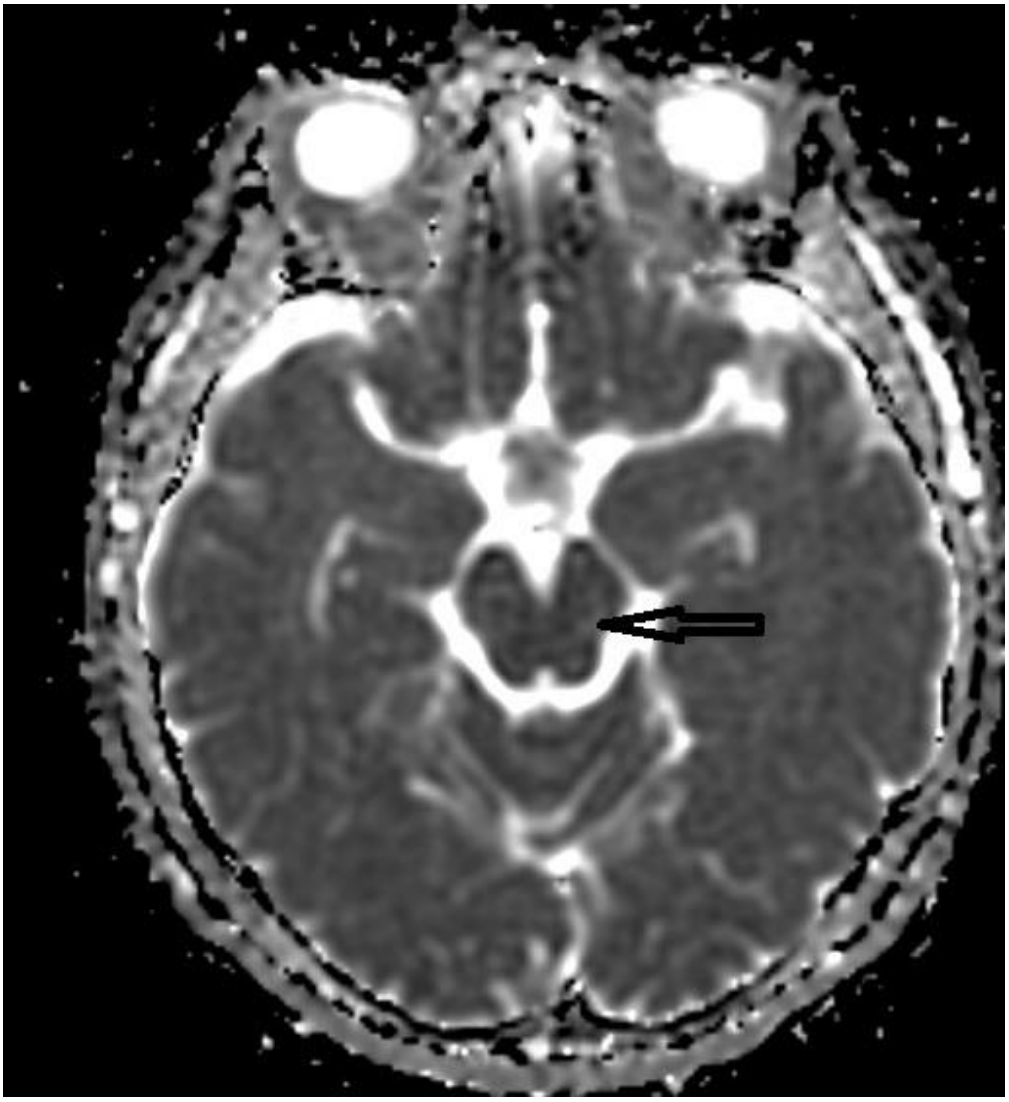
Description: The sagittal T2 FLAIR describes the involvement of hypothalamus and tuber cinereum (red arrow). This findings may explain the central diabetes insipidus seen in our patient. Note also the pontine lesions. **Origin:** Department of Radiology, San Paolo Hospital, Milan, Italy.

c



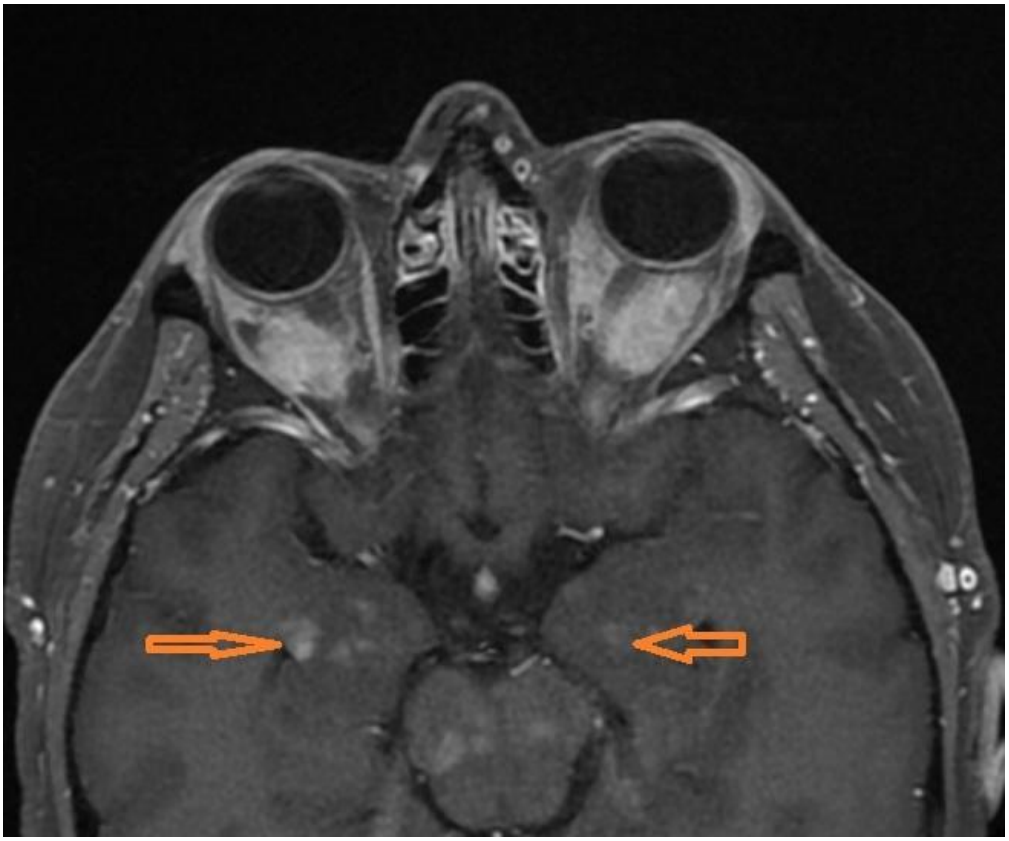
Description: The DWI showed a mild restriction of diffusion of the pontine lesions (black arrow). **Origin:** Department of Radiology, San Paolo, Milan, Italy.

d



Description: The corresponding ADC map does not show the presence of restricted diffusivity in the pons (black arrow). **Origin:** Department of Radiology, San Paolo Hospital, Milan, Italy.

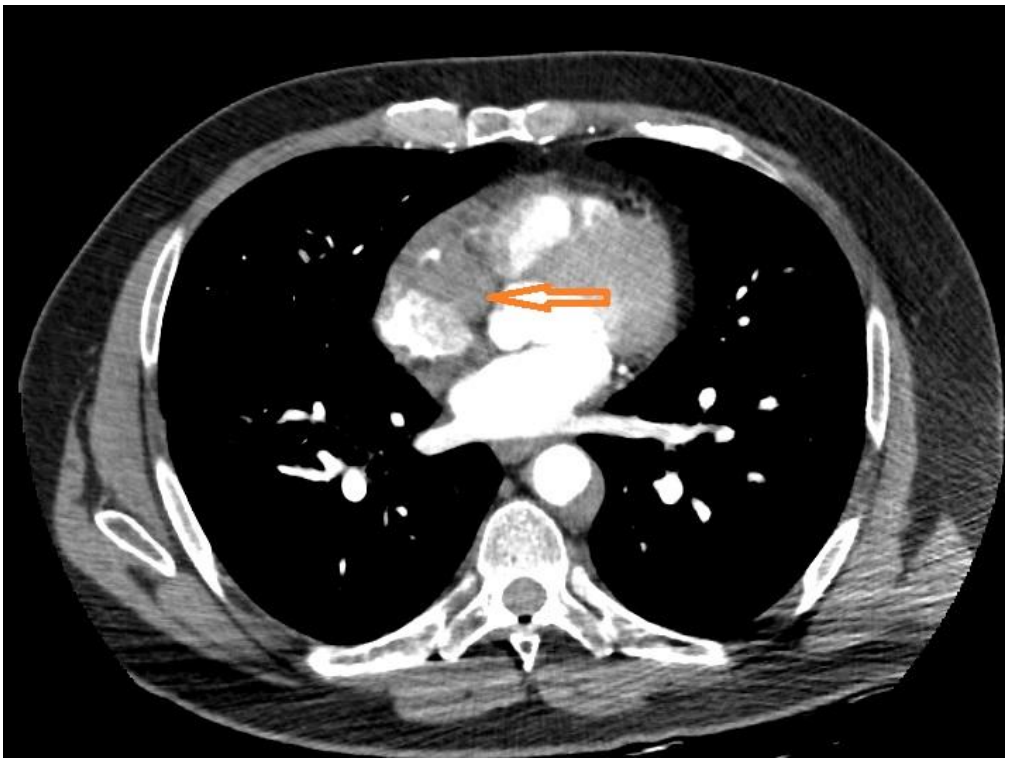
e



Description: This axial post-contrast T1 fast spin echo image reveals multiple, nodular enhancing lesions in the hippocampal (orange arrow) and pontine region. **Origin:** Departmwent of Radiology, San Paolo Hospital, Milan, Italy

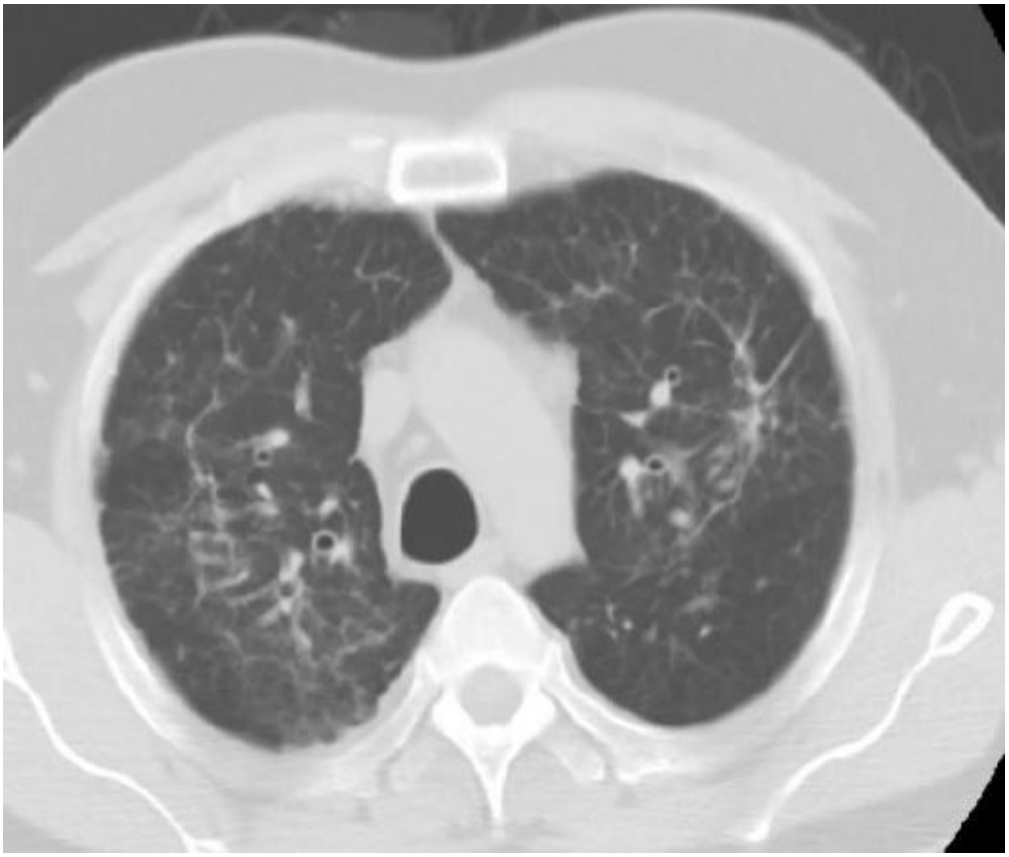
Figure 4

a



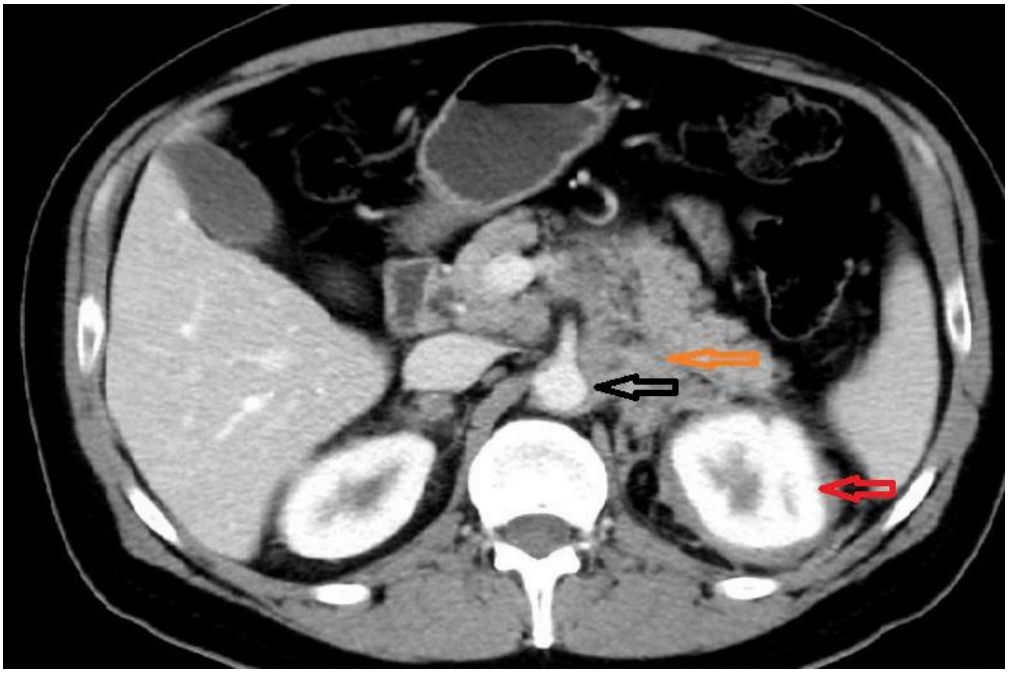
Description: The contrast-enhanced CT of the thorax demonstrates the presence of a fibrotic mass in the rigth periatrial fat (orange arrow). **Origin:** Department of Radiology, San Paolo Hospital, Milan, italy.

b



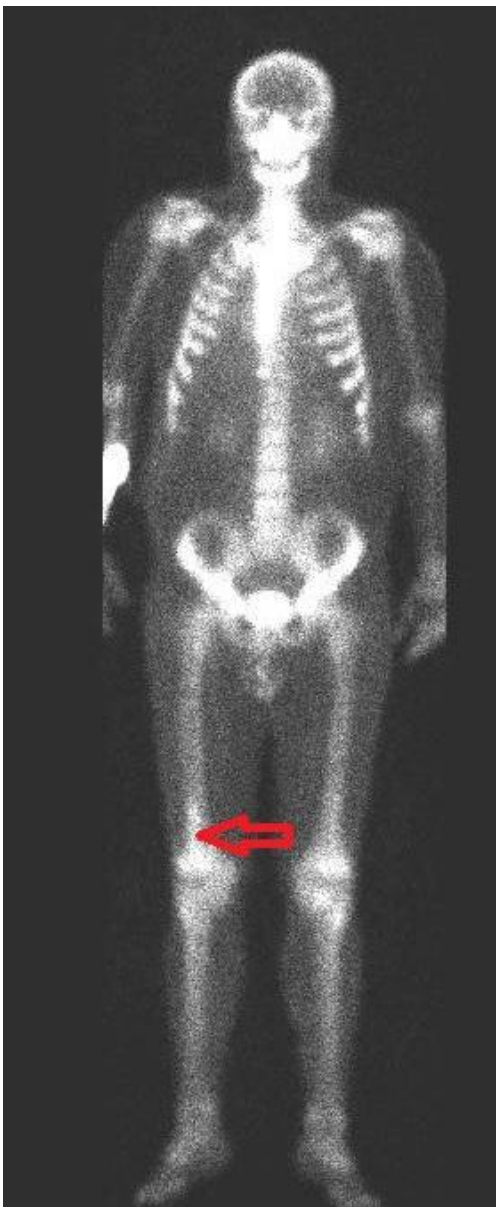
Description: An axial CT image of lungs shows the presence of thickened interlobular septa and ground glass areas in the upper lobes. **Origin:** Department of radiology, San Paolo Hospital, Milan, Italy

c



Description: A wide fibrotic mass in the retroperitoneum (orange arrow) with perirenal and periaortic involvement, which produces the hairy kidneys sign (red arrow) and the coated aorta sign (black arrow), respectively. **Origin:** Department of Radiology, San Paolo Hospital, Milan, Italy

d



Description: The technetium-99m bone scintigraphy describes a pathological uptake in the right distal femur (red arrow). **Origin:** Department of Radiology, San Paolo Hospital, Italy