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Erdheim-Chester Disease: MR of Intraaxial and Extraaxial Brain Stem Lesions

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Summary: A case of Erdheim-Chester disease demonstrates cerebral hemispheric involvement, as well as and intraaxial and extraaxial brain stem involvement in a patient with symptoms of paraparesis, urinary incontinence, visual loss, ataxia, vertigo, proptosis, and nystagmus. Persistent gadopentetate dimeglumine enhancement was noted in the extraaxial cervicomedullary brain stem lesion 23 days after injection. However, the supratentorial lesions fail to show similar persistent enhancement. This case also demonstrates MR features characteristic of retrobulbar infiltration.

Index terms: Brain stem, magnetic resonance; Histiocytosis

Erdheim-Chester disease is characterized by the presence of lipid-laden histiocytic infiltration of multiple body systems. A case with several unusual features is presented.

Case Report

A 66-year-old normotensive white woman with Erdheim-Chester disease, proved by biopsy in 1987, presented with a 2-year history of bilateral lower-extremity weakness, progressing to total paraparesis 2 weeks before admission. During the same period fecal and urinary incontinence also developed. Evaluation of the prehospitalization clinical course reveals a 15-year history of multiple neurologic symptoms consisting of visual loss, vertigo, and ataxia. Concomitant development of proptosis and nystagmus was also reported. Significant endocrine abnormalities were limited to hypothyroidism.

Physical examination revealed proptosis and xanthelasma in both upper and lower eyelids, associated with a milky discharge from both medial canthi. No adenopathy or joint pain was noted. Marked lower-extremity weakness was noted. Neurologic evaluation revealed decreased visual and auditory acuity. Nystagmus was elicited on extreme right lateral gaze. Although gross ocular movements were intact, inability to close the left eye was demonstrated, secondary to involvement of the orbicularis oculi muscle with multiple xanthelasma.

Radiography of the appendicular skeleton revealed multiple foci of medullary sclerosis. In the lower extremi-

ties, the sclerotic changes extended to the subarticular surface of the long bones. No definite lytic lesions were seen. The flat bones were unremarkable.

Evaluation of the brain was performed using 1.5-T magnetic resonance (MR) imaging, on July 8, 1994. T1-weighted (500/20/1 [repetition time/echo time/excitations]) images were obtained before the administration of gadopentetate dimeglumine. Postcontrast T1-weighted images were also obtained. Multiple cerebral (Fig 1A) and pontine (Fig 1B) lesions were demonstrated, as well as a large extraaxial mass at the level of the foramen magnum, which is hyperintense on all precontrast T1 images (Fig 1C). Comparison with an outside study of June 13, 1994, clearly shows the lesion to have been hypointense on precontrast images (Fig 1D). It was only after the administration of gadopentetate dimeglumine that marked hyperintensity was noted on this prior study (Fig 1E). Therefore, the high signal noted within this extraaxial lesion on the precontrast T1-weighted images of July 8, 1994 is the result of persistent gadolinium retention from the study of June 13.

Evaluation of the orbits reveals the presence of bilateral bulky infiltrating masses surrounding the optic nerves (Fig 1F). Associated findings include atrophy of the extraocular muscles and the right optic nerve.

During the present hospitalization, fractionated radiation therapy was used in an attempt to diminish the size of the posterior fossa mass, with disappointing clinical results. Prior radiation therapy had also been used to treat the initial manifestations of retrobulbar disease, with limited success.

Discussion

Since 1930, when two cases of Erdheim-Chester disease were described, a great deal of controversy has surrounded the pathogenesis and classification of this entity. Many authors have suggested that Erdheim-Chester disease is part of the spectrum of histiocytosis X (1-3), whereas others believe that there are sufficient cellular ultrastructural differences to classify

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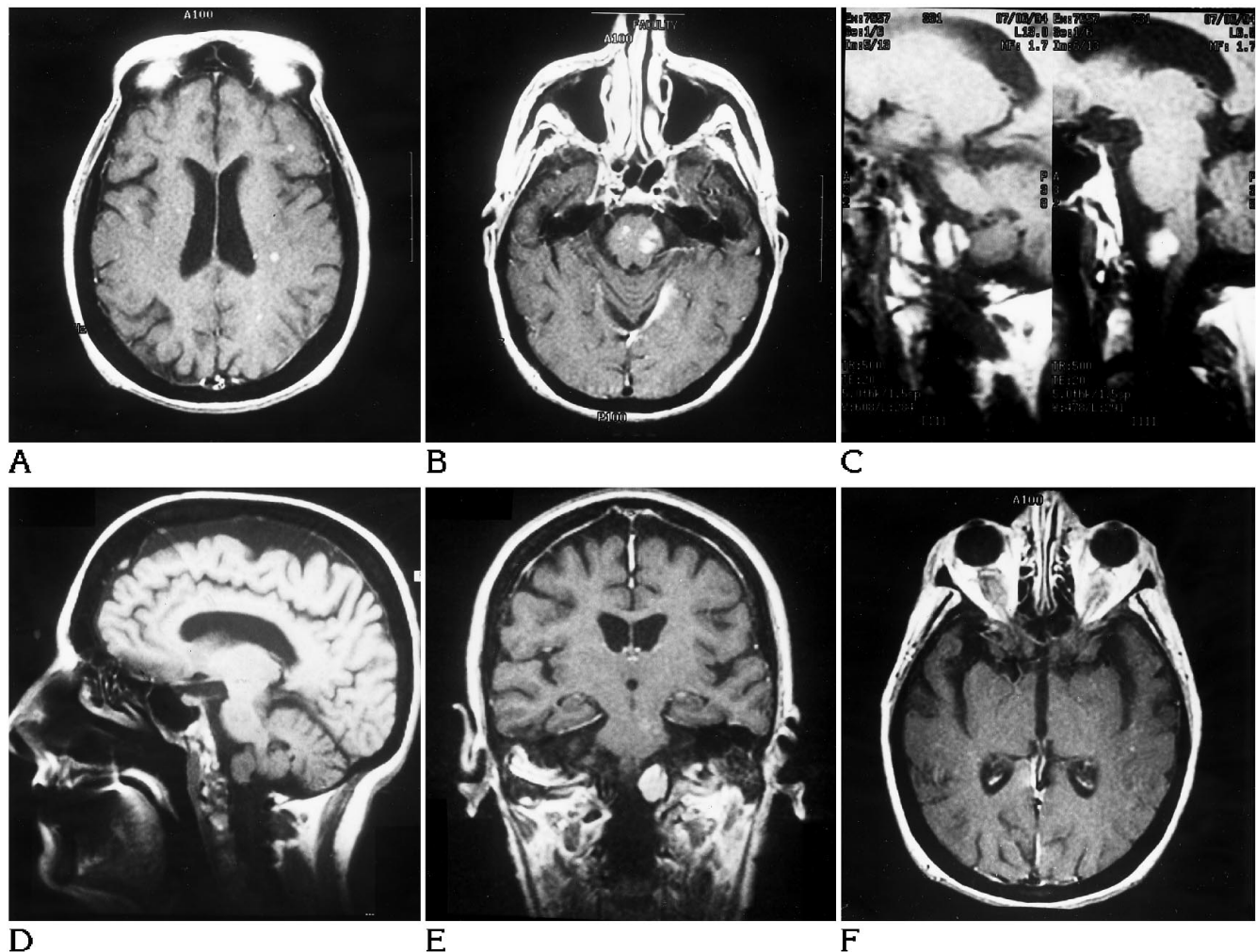


Fig 1. A, Contrast-enhanced axial T1-weighted MR image shows enhancement of white matter lesions bilaterally. These lesions were isointense to white matter on precontrast images.

B, Contrast-enhanced axial T1-weighted MR image shows meningeal enhancement and multiple enhancing pontine lesions that were imperceptible on pre-gadopentetate dimeglumine images. Note their location within the region of the corticospinal tracts, bilaterally.

C-E, Histiocytic lesion causing medullary compression. C, Precontrast T1-weighted sagittal MR image reveals a hyperintense extraaxial lesion causing "kinking" of the brain stem. The marked intensity of this lesion is caused by persistent gadolinium retention from the previous study (performed 23 days before present study). Prior outside T1-weighted sagittal precontrast (D) and coronal postcontrast (E) MR images show intense enhancement of the left extraaxial brain stem lesion. This study was performed 23 days before the present study.

F, Axial postcontrast T1-weighted MR image shows enhancement of bilateral bulky retrobulbar masses associated with atrophy of extraocular muscles and the right optic nerve.

Erdheim-Chester disease as a distinct lipoidosis (4, 5). The unique nature of Erdheim-Chester disease is supported by both electron microscopy and immunoperoxidase studies (4, 6).

Clinically, Erdheim-Chester disease has a male predominance (2.7:1), affecting mostly white patients (5). The median age is approximately 56.5 years, although patients as young as 26 years and as old as 78 years have been reported (1). Patients are usually asymptomatic

(42%) (1). The most common presenting symptom is bone pain (32.5%), and 32% of patients report fever and weight loss (1). Physical examination reveals no consistent findings; however, hypertension has commonly been noted, and adenopathy has not been reported (5). Laboratory evaluation also tends to reveal no remarkable findings; however, abnormal levels of neutral fat, free fatty acids, and triglycerides have been reported (1).

The natural history of the disease ranges from limited and localized to systemic and fatal. Shields et al (7) reported cardiomyopathy, severe lung disease, and chronic renal failure as main causes of death. However, the appendicular skeletal system is the most commonly affected. Plain-film radiographic findings consist of bilateral, symmetric metadiaphyseal sclerotic lesions of the long bones, with sparing of the epiphyses and joint space.

Central nervous system involvement is rare, except for posterior pituitary and retroorbital involvement. The latter ranges from subtle perineural infiltration to diffuse homogenous masses associated with proptosis and extraocular muscle atrophy (7, 8). Intense postcontrast retroorbital enhancement is usually observed with both computed tomography and MR (7). Posterior pituitary involvement has been elegantly described by Tien et al (9). In the presence of diabetes insipidus, secondary to infiltration by Erdheim-Chester histiocytes, hypointensity of the posterior lobe is noted on T1-weighted images (9). In the absence of diabetes insipidus, the normally observed hyperintense T1 signal is present (9).

Cerebral intraparenchymal involvement has been reported in one other patient (10). The present case not only shows bilateral enhancing deep white matter cerebral lesions but also demonstrates the presence of intraaxial pontine lesions and extraaxial brain stem involvement. Evaluation of the latter lesion reveals precontrast T1 hyperintensity, secondary to persistent gadolinium retention (Fig 1C). Tien et al (10) previously described this phenomenon in a patient with multiple intraparenchymal cerebral lesions. Although the exact mechanism of persistent enhancement has not been elucidated, findings suggest that gadopentetate dimeglumine or a related metabolite may be retained by these unique histiocytes (10). However, the present case fails to reveal similar retention in any supratentorial lesion. Therefore, it is evident that although such retention is seen only in Erdheim-Chester histiocytic lesions, it is not a universally observed feature, nor is it limited only to intraparenchymal lesions.

Unlike the patient's visual symptoms, the remaining clinical findings cannot be as easily accounted for, based solely on the MR findings. Given the patient's history of lower-extremity weakness, progressing to paraparesis, an MR

spinal survey was performed at an outside institution, without gadolinium. This study failed to reveal any focal lesions, and, unfortunately, further spinal evaluation was unable to be performed during the present admission. However, the presence of multiple lesions in the region of the pontine portion of the corticospinal tracts (Fig 1B), as well as the presence of the large extraaxial brain stem mass causing medullary compression (Fig 1C), might contribute to the clinical findings.

To date, no one treatment has proved to be universally efficacious. Lantz et al (5) described the use of nonsteroidal antiinflammatory drugs (indomethacin), corticosteroids (prednisone), and antineoplastic drugs (vinblastine) for the treatment of diffuse bone lesions. Unfortunately, each of these regimens has met with limited success. In some patients with a suboptimal response to chemotherapy, fractionated radiation therapy has yielded positive results (1, 5). The role of radiation therapy for the treatment of central nervous system involvement has not been completely evaluated. As stated previously, no significant palliation of symptoms or diminution of posterior fossa masses was observed in the present case.

The differential diagnosis for the spectrum of intracranial disease found in Erdheim-Chester disease included granulomatous disorders, such as sarcoidosis, and idiopathic disorders, such as histiocytosis X. Each of these disorders may cause optic nerve and posterior pituitary involvement, as well as dural thickening and enhancing parenchymal nodules. However, persistent gadolinium enhancement is a unique distinguishing feature of Erdheim-Chester disease.

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