A Rare Condition of Hand-Schüller-Christian Disease

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The authors present the clinical case of a 61-year-old patient with Hand-Schüller-Christian disease associated with multisystemic involvement. The onset of such puzzling symptoms and the extremely rarity of this disease in a patient of such advanced age resulted in a delayed diagnosis and subsequently delayed treatment of the patient.

Key Words: Hand-Schüller-Christian, X-cell histiocytosis, eosinophilic granuloma.

X-cell histiocytosis is a nosologic entity discovered by Lichtenstein in 1953.[1] This pathology presents three clinical symptoms that are expressions of the same disease but that are different in symptoms and evolution, with a localized, spread acute, or chronic evolution: the eosinophilic granuloma, the Hand-Schüller-Christian disease, and the Letter-Siwe disease.[1a,2] The most appropriate name for this disease is Langherans-cell histiocytosis as the cellular disorder seems to be attributable to the proliferation and subsequent dissemination of a large number of this type of cell in several tissue types. Histiocyte proliferation, often having a granulomatous aspect, is the chief histological feature resulting from several clinical pictures.[1a]

Currently its incidence is estimated to be approximately 1:2,000,000 per year.[1a] Eosinophilic granuloma represents the most benign and localized form; in the majority of cases it affects young adults and occurs mainly in the cranial and facial nerves.

Letter-Siwe disease, being a systemic acute form, affects mostly pediatric patients and, because of the severe damage to the bones and viscera, the prognosis is fatal in the majority of cases.

The spread chronic form, Hand-Schüller-Christian disease, is typically characterized by the following symptoms: exophthalmos, osteolytic lesions, and because of the frequent involvement of the pituitary gland, diabetes insipidus. Rarely, however, does a patient exhibit the entire triad at once.[1a] At the systemic level, hepatic, splenic, bone, pulmonary, gastrointestinal, lymphonodal, and central nerve system granulomas are reported. Moreover, there may be skin lesions provoked by hemorrhagic events or lesions which may simulate seborrhoic dermatitis.[3]

The craniomaxillofacial bones are frequently involved and present osteolytic lesions of the mandible, the superior maxillary, and above all the petrous pyramid of the temporal bone with subsequent onset of otorrhea. The involvement of the oral cavity results in floating teeth, pain, maxillary tumefactions, oral cavity ulcerations, and periodontal lesions; and it may often represent the first clinical manifestation of the disease.

The authors present the case of a 61-year-old female patient with Hand-Schüller-Christian disease where the right diagnosis could have been influenced by the extraordinary presence of such a disease in a patient of such advanced age.

Case Report

The 61-year old woman was examined for bilateral pain in the upper and lower jaw.

Clinical History

Because of widespread arthralgias, pelvic and spinal radiographic examination was performed 3

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years earlier. The examination highlighted the presence of multiple osteolytic areas.

During a hospitalization in April 1998, she underwent echoabdomen, which detected splenomegaly and a slight dishomogeneity caused by steatosis in the liver; the medullar biopsy elucidated lack of neoplastic cells.

During a successive hospitalization in June 1998, the patient underwent total body a computed tomography (CT) scan, which documented "osteostructural alterations within the left scapula (spinous process and posterior arch), the IV left rib, and wide osteolytic areas within the left iliac wing and the pubic symphysis . . .".

In October 1998, the patient underwent a series of instrumental investigations:

1. bone scintigraphy of the right occipital wall and of the distal part of the tibia bilaterally, which detected a hypercaptation within the left maxillary region;
2. facial nuclear magnetic resonance (NMR), which showed a bone lesion within the sphenoid and the occipital basis as well as the presence of tissutal quota within the left mandibular region.

In November 1998, the patient was hospitalized at an orthopedic clinic, where she underwent a biopsy of the osteolytic lesions (right tibia and pubic symphysis) which showed partially necrotic lamellar bone tissue surrounded by fibrous tissue with foci of filtered lymphomononucleates and several neutrophile granulocytes such as found in chronic osteomyelitis. This led to the patient's undergoing antibiotic therapy. In January 1999, she underwent follow-up bone scintigraphy showing the persistence of hypercaptative areas (within the medial region of the mandible, the left maxilla, the tibia bilaterally, the left sacroiliac region, the left scapula, the distal extremity of the femur bilaterally, and a new area within the right mastoid region.

Clinical Assessment

The objective examination of the craniofacial district elucidated the presence of cutaneous eruptions with a xanthomatous aspect in the paralateronasal region bilaterally and right otorrhea with a purulent aspect; following the endo-oral inspection, the presence of all dental elements was reported together with a painful tumefaction in the left parasinphisarial maxillary region. A CT scan of the face was requested and performed in September 2000. It documented a farther evolution of the bone lesions,

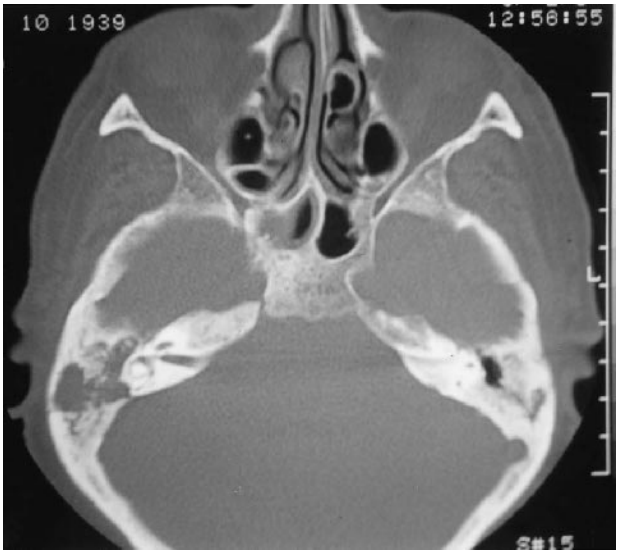

Fig 1 CT scan: osteolytic lesions in the mastoid area

with "osteolytic areas in the right mastoid; destruction of the external wall of the temporal pyramid; and thickening of the adjacent soft tissues, medial ear cavity, mastoid cells, and sphenoid cavities opacity. These symptoms were compatible with chronic phlogosis. Areas of widespread osteolysis were found in the maxilla and lingual body, being of larger volume in the simphisarial region of the mandible" (Figs 1, 2, 3).

This led to the patient's undergoing surgical cu-

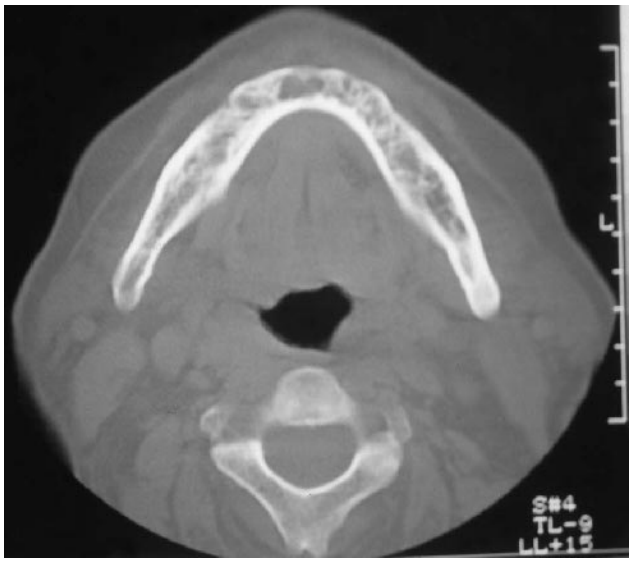

Fig 2 CT scan: osteolytic lesions in the simphisarial region of the mandible

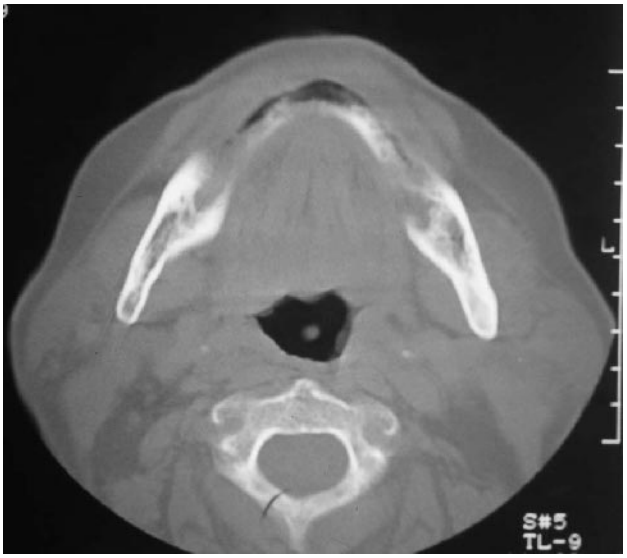

Fig 3 CT scan: intraosseous mandibular lesions

rettage and biopsy of a tissue portion that extended to the mastoid cavern. The histological examination revealed the presence of Langerhans-like histiocyte cells associated with eosinophilic and neutrophilic disorders and hyperplastic blood vessels.

All the clinical, radiologic, and histologic data allowed the diagnosis of Hand-Schüller-Christian disease. The patient is still undergoing a proper pharmacologic treatment with administration of Vinblastine: 10 mg/w and Prednisone: 50 mg/d.

Discussions and Conclusions

Diagnosis of X histiocytosis is not always easy because of the high variability of its signs and symptoms at the onset of the disease. The delayed diagnosis in this patient is due to the nonspecificity of initial symptoms but especially because of the extremely rare onset of the disease in a patient of advanced age. The typical three symptoms including exophthalmos, diabetes insipidus, and osteolytic lesions are rare at the onset of this pathology; however, this does not invalidate the correct diagnostic approach.

The patient we observed exhibited neither exophthalmos nor diabetes insipidus, but along with the presence of bone lesions, there were other signs compatible with X histiocytosis, although they were not pathognomonic, such as xanthomatous lesions of the face, splenomegaly, and hepatic pain. Otorrhea with osteolytic lesions of the mastoid was more revealing along with other osteolytic-like lesions such as those that were reported in the face and in the long bones (Fig 1). The study of the outcomes of the CT examination of the craniofacial district raised doubt about the presence of a systemic pathology involving the bones and the soft tissues because of the peculiar distribution of the lesion (Figs 2, 3). The precautionary approach of the diagnosis resulted from the extreme rarity of such pathology, especially in patients aged more than 50 years. In 1978, Dolezal and Thomson discussed a 76-year-old patient with Hand-Schüller-Christian disease and reviewed the current international literature in the English language and verified that this pathology affected 76% of those aged less than 10 years and 91% of patients by age 30 years. Only in 4% of the cases was the onset of this disease reported after the fifth decade of life, while it was even more rare in successive decades.[3] It is important to remember that this pathology, although it occurs in the first decade of life in the majority of cases, may actually affect any age, and only a correct clinical and radiological diagnosis followed by bioptical confirmation avoids incidental therapeutic mistakes.

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