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Case Report

Systemic mastocytosis – a diagnostic challenge

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ABSTRACT

Mastocytosis refers to a group of disorders characterized by the infiltration of clonally derived mast cells to the skin or extracutaneous tissues resulting in a heterogeneous clinical picture. It is a rare hematologic disorder in all its forms. The exact incidence is unknown; it affects patients of any age and males and females equally. Its molecular pathogenesis is incompletely understood. The clinical features of mastocytosis result from both chronic and episodic mast cell mediator release, signs and symptoms arising from diffuse or focal tissue infiltration, and, occasionally, the presence of an associated non-mast cell clonal hematologic disease. The histopathologic analysis is essential for definitive diagnosis but there is no curative treatment. The authors report a clinical case of a 72-year-old woman with no history of allergies, with bicytopenia, weight loss, and diffuse axial osteolytic lesions. This is a rare clinical case of aggressive systemic mastocytosis for which palliative treatment can improve survival and quality of life. A brief review of the literature about this pathology is also included.

Introduction

Mastocytosis refers to a group of myeloproliferative disorders characterized by an excessive proliferation of mast cells and their accumulation in one or multiple tissues. According to the World Health Organization (WHO), the disease can be classified as cutaneous mastocytosis (CM), which describes forms of mastocytosis that are limited to the skin, and systemic mastocytosis (SM) in which mast cells infiltrate extracutaneous organs, with or without skin involvement.[1,2] The pathogenesis is not well defined and treatment is only on a palliative basis.

The authors report a clinical case of SM and briefly review the literature about this disease.

Clinical case

A 72-year-old Caucasian woman with a history of diverticulosis, surgically-corrected lumbar hernia and plurisegmental degenerative osteoarticular disease is reported. There was

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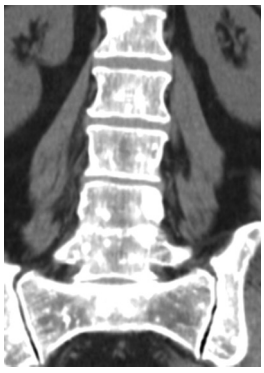

Figure 1 – Computed tomography scan of the lumbar spine and sacrum – scattered osteosclerotic lesions on vertebrae, sacrum and iliac bones (coronal view).

no background of allergies, smoking or alcoholism and she was taking only analgesic medications. In May 2012, she was admitted for acute diverticulitis. During her hospital stay the lower back pain worsened and so a computed tomography (CT) scan of the lumbar spine was performed which documented diffuse osteopenia and multiple scattered osteosclerotic lesions on vertebrae, the sacrum and the iliac bones (Figure 1). After discharge and with a presumptive diagnosis of occult neoplastic disorder, she was referred to her doctor for further studies.

Several medical tests were performed including blood analysis, endoscopic and imaging exams but none showed relevant changes. In September she started with persistent diaphoresis with no fever or any symptoms suggestive of an infectious focus, anorexia and a quantified weight loss. The blood tests revealed slightly elevated levels of alkaline phosphatase (162 U/L) and lactate dehydrogenase (LDH – 454 U/L). She was admitted again and a CT body scan was performed (Figure 2) which showed osteolytic lesions, in addition to the diffuse osteosclerotic lesions previously documented, without expansive features, spread throughout the axial skeleton, that were assumed to be bone marrow sclerosis phenomena. Diaphoresis was associated to a pharmacological iatrogenic effect and the weight loss to a reactive depression. She was discharged and referred to our Internal Medicine Department.

The sequential control blood tests showed increasing levels of bicytopenia (hemoglobin 11.2 g/dL and platelet count 122×10^9/L), leukocytosis (19.40 x 10^9/L), with no formula inversion, and LDH (572 U/L). With strong suspicion of a hematological disorder, a blood smear, myelogram and bone marrow biopsy were performed but all were unrevealing. The immunological study was negative and Paget's disease and multiple myeloma were excluded. Finally, a percutaneous L1 biopsy was performed. This was essential for the definitive diagnosis because it documented a multifocal infiltration of atypical mast cells, characterized by spindle-shaped and hypogranular forms, representing 5% of cellularity and forming cellular aggregates of more than 15 cells. An immunohistochemistry study identified positive staining for CD117 and tryptase of these mast cells confirming the diagnosis of SM (Figures 3–5). These features associated with peripheral blood test abnormalities (bicytopenia), weight loss

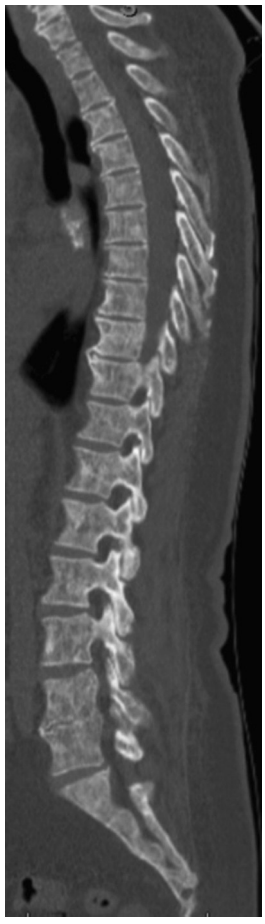

Figure 2 – Computed tomography scan of the lumbar spine and sacrum – osteosclerotic and osteolytic lesions, without expansive features, spread throughout the axial skeleton (sagittal view).

and presence of osteolytic lesions are sufficient C findings that allowed the classification of an aggressive form of SM.

The patient was referred to the Hematology Department and began chemotherapy with cladribine which gave a

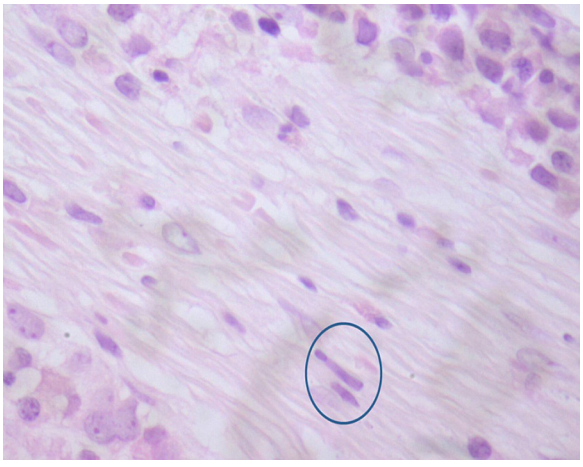

Figure 3 – Hematoxylin-eosin stain (magnification: 400×) – peripheral fibrosis and atypical mast cells with fusiform shape (circle).