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Case Report

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SYSTEMIC MASTOCYTOSIS WITH AN ASSOCIATED MYELOPROLIFERATIVE NEOPLASM: CASE REPORT

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ABSTRACT

Systemic mastocytosis is a rare disease. We describe a new case of systemic mastocytosis with an associated hematologic neoplasm. A 36-year-old woman hospitalized for exploration of febrile asthenia and bone pain. The chest X-ray showed a heterogeneous appearance of the sternum with mixed osteocondensing and osteolytic process. The bone scintigraphy revealed several foci of hyperfixation in the sternum, humerus, femurs and pelvis. The myelogram as well as the bone marrow biopsy revealed rounded cells with a central nucleus and a finely granular, eosinophilic cytoplasm. This form of systemic mastocytosis with an associated hematologic neoplasm is rarely reported.

KEYWORDS: Systemic mastocytosis, myeloproliferative neoplasm.

INTRODUCTION

After the World Health Organization (WHO) classification of myeloid neoplasms revesion in the 2106, mastocytosis is no longer considered a subgroup of myeloproliferative neoplasms (MPN), but is considered a distinct disease category.[1,2]

Its clinical presentation is heterogeneous, ranging from skin-limited disease (cutaneous mastocytosis), particularly in pediatric patients,[3–6] to a more aggressive forms with extra-cutaneous involvement with a systemic mastocytosis (SM) that may be associated with multiorgan dysfunction and shortened survival and which is generally seen in adult patients.[7]

We describe a new case of systemic mastocytosis with an associated hematologic neoplasm.

CASE REPORT

36-year-old woman hospitalized for exploration of febrile asthenia and bone pain. The patient had fever and the clinical examination of the lymph nodes and abdominal areas was without abnormalities. All the joints were free. Biology revealed an inflammatory syndrome (VS at 124 mm; CRP: 54 mg/L). The hemoglobin was at 8.8 g/dL, white blood cells at 5270/mm3 and platelets: 174,000/mm3. The chest X-ray showed a heterogeneous appearance of the sternum with mixed osteocondensing and osteolytic process. The bone scintigraphy revealed several foci of hyperfixation in the sternum, humerus, femurs and pelvis (figure 1). Blood disease was suggested and the myelogram as well as the bone marrow biopsy revealed rounded cells with a central nucleus and a finely granular, eosinophilic cytoplasm. These cells expressed CD68 and C-Kit immunohistochemistry, confirming their mast cell nature. In addition, megakaryocytic abnormalities were noted in favor of an associated chronic myeloproliferative syndrome. The search for other visceral sites of mastocytosis by abdominal CT scan was negative. The bone densitometry was normal. The diagnosis of aggressive systemic mastocytosis was retained.

Oral corticosteroids (1 mg kg/day of prednisone) were started. At the same time, treatment with interferon alfa 3 million units, 3 times per week, was initiated due to the aggressive and systemic nature (bone marrow involvement). Pain in the lower limbs was successfully treated with an opioid drug. After six months of treatment, the general condition improved, the patient regained her walking autonomy and the inflammatory syndrome resolved. Monitoring of clinical status and biology was regular. The patient had a pregnancy under corticosteroid therapy at a dose of 10 mg/day with good progress. A year later she was seen again for acute febrile respiratory failure. The x-ray showed bilateral alveolo-interstial syndrome with the presence of lymphopenia at 700elt/ml. The search for mycobacterium tuberculosis came back positive and the patient was put on anti-tuberculosis treatment for 6 months with good clinical outcome and 1 year follow-up.

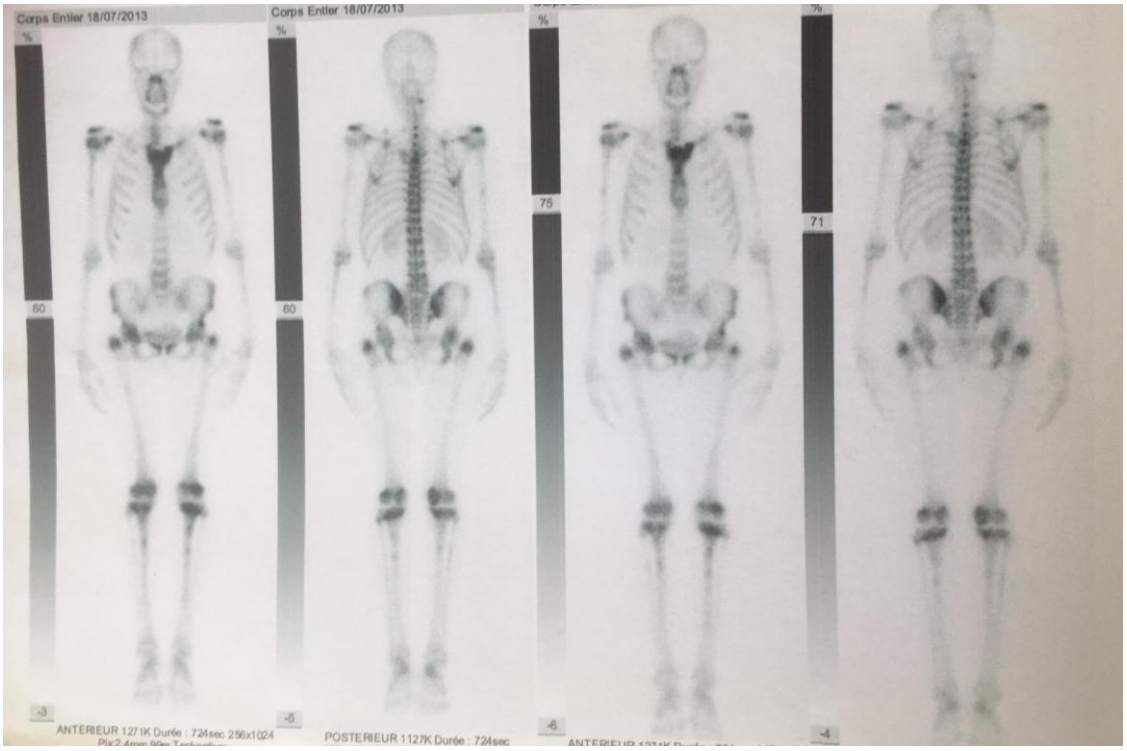

Figure 1: Bone scintigraphy revealing several foci of hyperfixation in the sternum, humerus, femurs and pelvis.

DISCUSSION

There are five subtypes defined include: indolent systemic mastocytosis (ISM), smoldering systemic mastocytosis (SSM), aggressive systemic mastocytosis (ASM), systemic mastocytosis with an associated hematologic neoplasm (SM-AHN), and mast cell leukemia (MCL).[8] Both ISM and SSM have relatively good prognoses but still with many quality of life issues related to symptoms.[9] our patient had SM-AHN form with good outcomes under treatment.

In practice, the current diagnostic approach for SM starts with a bone marrow (BM) examination as this site is generally involved in adult mastocytosis, and histological diagnostic criteria for non-BM, extracutaneous organ involvement in SM are not standardized. Further, BM examination also allows detection of a second hematologic neoplasm, if present.[10,11] In our case, BM biopsy showed a chronic myeloproliferative syndrome.

SM-AHN is a diagnosis involving meeting both the criteria for systemic mastocytosis while additionally the criteria for another associated disorder such as myeloproliferative neoplasm, myelodysplastic syndrome, or lymphoproliferative disorder.

SM-AHN tends to be observed in older patients with a more aggressive clinical course.[12] Types of associated haematological malignancies included chronic myelomonocytic leukaemia, myelodysplastic syndrome, B-cell lymphoma/leukaemia/plasma cell neoplasms, myeloproliferative neoplasms and acute myeloid leukaemia.[13]

The treatment of adult SM is highly individualized; recent data reveal encouraging clinical and histologic responses following treatment with small molecule kinase inhibitors that potently target activation loop mutants of KIT receptor, cementing the view that KITD816V represents the driver mutation for SM.[14-16]

Interferon-α (IFN-α) has proved clinical activity in symptomatic SM. since the initial report in 1992,101 several case reports or small series have shown its ability to improve symptoms of MC degranulation and decrease BM infiltration.[17,18] our patient had well out comes under IFN- α.

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