

Systemic mastocytosis

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Section: Musculoskeletal system

Area of Interest: Bones

Procedure: Education

Imaging Technique: CT

Special Focus: Pathology Haematologic diseases

Case Type: Clinical Cases

Authors: Miguel Nogueira¹, Inês Rolla¹, Daniel Mer²,
Ana Villanueva², Elena Utrera², Iosu Antón Badiola³,
Francisco Tardáguila Montero²

Patient: 66 years, female

Clinical History:

Asymptomatic 64-year-old male patient. Routine chest X-ray is performed.

Imaging Findings:

Chest X-ray shows diffuse sclerosis of the humerus, ribs and thoracic vertebrae (Fig. 1). On the abdominal X-ray diffuse sclerosis of the lumbar vertebrae, sacrum, iliac and femoral heads could also be noted (Fig. 2).

Thoraco-abdomino-pelvic CT was then performed and the findings already seen at the plain radiographic films were confirmed (Fig. 3-4).

Bone biopsy confirmed the diagnosis of systemic mastocytosis (Fig. 5).

Discussion:

Mastocytosis is a myeloid neoplastic disease characterized by abnormal mast cell proliferation. Two major forms of this disease have been described: cutaneous mastocytosis and systemic mastocytosis (SM), which is further subdivided in four subtypes; (1) indolent SM, (2) SM associated with non-mast cell clonal haematological disease, (3) aggressive SM, and finally (4) mast cell leukaemia [1].

Clinical manifestations of SM may include anaemia, abdominal pain, diarrhoea, nausea, pruritus and cutaneous flushing. On physical examination hepatomegaly, splenomegaly and lymphadenopathy may also be present. The major diagnostic criterion for SM is the presence of dense infiltrates of mast cells in the bone marrow. Measurement of serum tryptase is also useful for the diagnosis, with values higher than 11.5 ng/mL found in more than 50% of the cases [2].

Although mastocytosis is mainly a clinical and laboratory diagnosis, it is important to recognize the radiographic features as radiologists may be the first suggesting the diagnosis.

Skeletal involvement is common. Bone abnormalities may be lytic, sclerotic or mixed, usually diffuse sclerotic involvement being the most commonly found. [3] The differential diagnosis of diffuse bone sclerosis is broad and includes fluorosis, haematological causes such as myelofibrosis, sclerosing bone dysplasias as osteopetrosis and pycnodysostosis and neoplastic causes as diffuse lymphoma, prostate or breast cancer metastases. Common abdominal imaging findings are quite nonspecific and include hepatosplenomegaly, retroperitoneal adenopathy, periportal adenopathy, mesenteric adenopathy and omental thickening.

The prognosis of mastocytosis is variable. Patients with cutaneous mastocytosis and indolent systemic mastocytosis remain stable for many years while patients with aggressive SM or mast cell leukaemia progress rapidly, usually

with a fatal outcome. [4]

Differential Diagnosis List: Systemic mastocytosis, Myelofibrosis, Metastatic carcinoma, Mastocytosis, Fluorosis, Pyknodysostosis, Osteopetrosis

Final Diagnosis: Systemic mastocytosis

References:

Akin C, Meltcalfe DD (2004) Systemic mastocytosis. Annu Rev Med 55:419-32. (PMID: [14746529](#))

Schwartz LB, Irani AM. (2000) Serum tryptase and the laboratory diagnosis of systemic mastocytosis. Hematol Oncol Clin North Am 14(3):641-57 (PMID: [10909044](#))

Johansson C, Roupe G, Lindstedt G et-al. (1996) Bone density, bone markers and bone radiological features in mastocytosis. Age Ageing 25 (1): 1-7 (PMID: [8670521](#))

Horny H, Sotlar K, Valent P (2007) Mastocytosis: State of the Art. Pathobiology 74:121-132 (PMID: [17587883](#))

Figure 1

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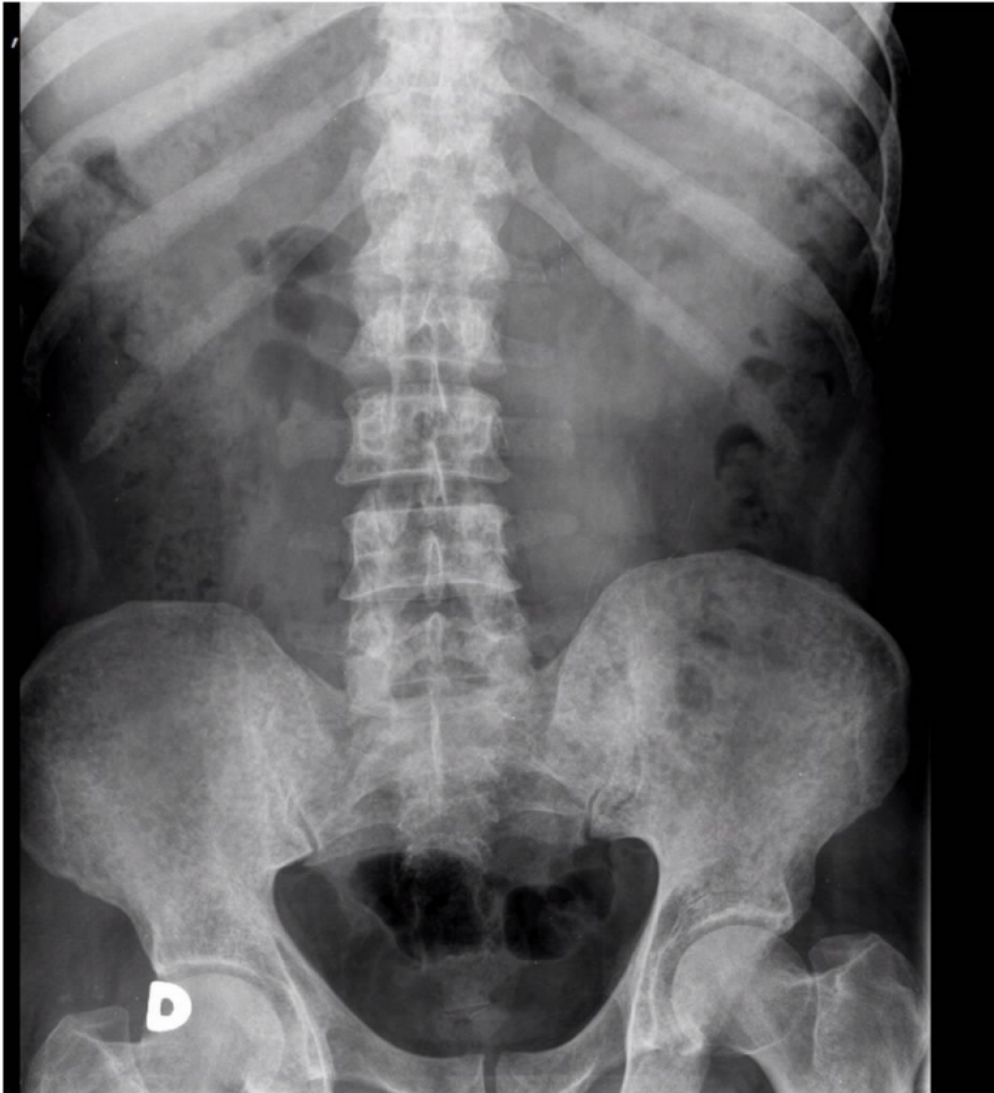


Description: Routine chest X-ray shows diffuse sclerosis of the humerus, ribs and thoracic vertebrae.

Origin: Radiology Department, Hospital Povisa, Vigo, Spain

Figure 2

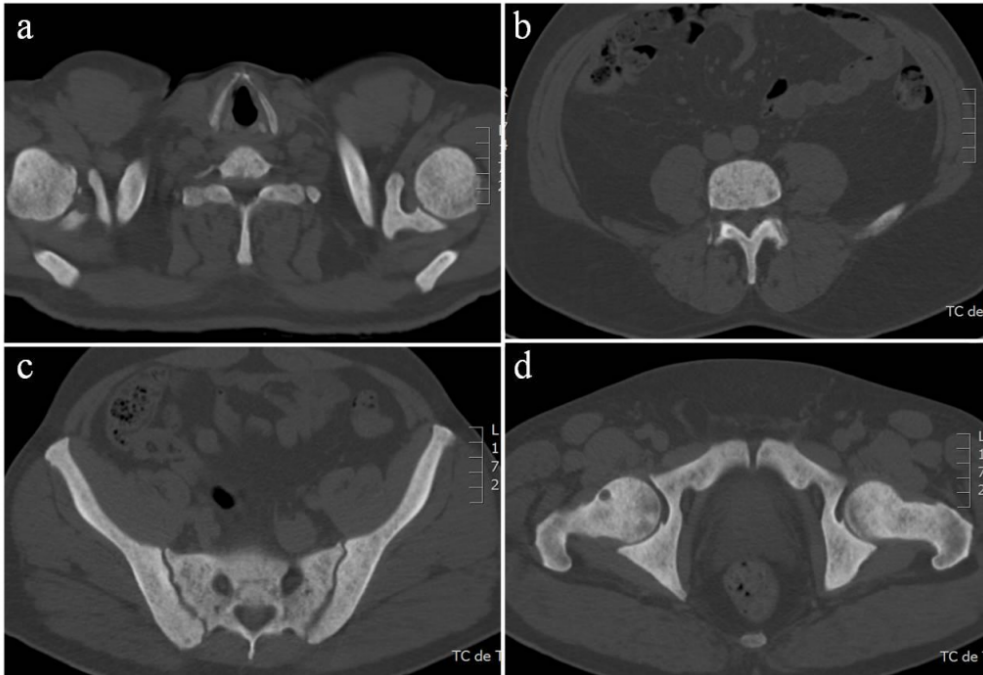
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Description: Abdominal X-ray showed diffuse sclerosis of the lumbar vertebrae, sacrum, iliac and femoral heads. **Origin:** Radiology Department, Hospital Povisa, Vigo, Spain

Figure 3

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Description: CT scan shows the same findings already noted previously at the plain radiographic studies: diffuse bone sclerosis. **Origin:** Radiology Department, Hospital Povisa, Vigo, Spain

Figure 4

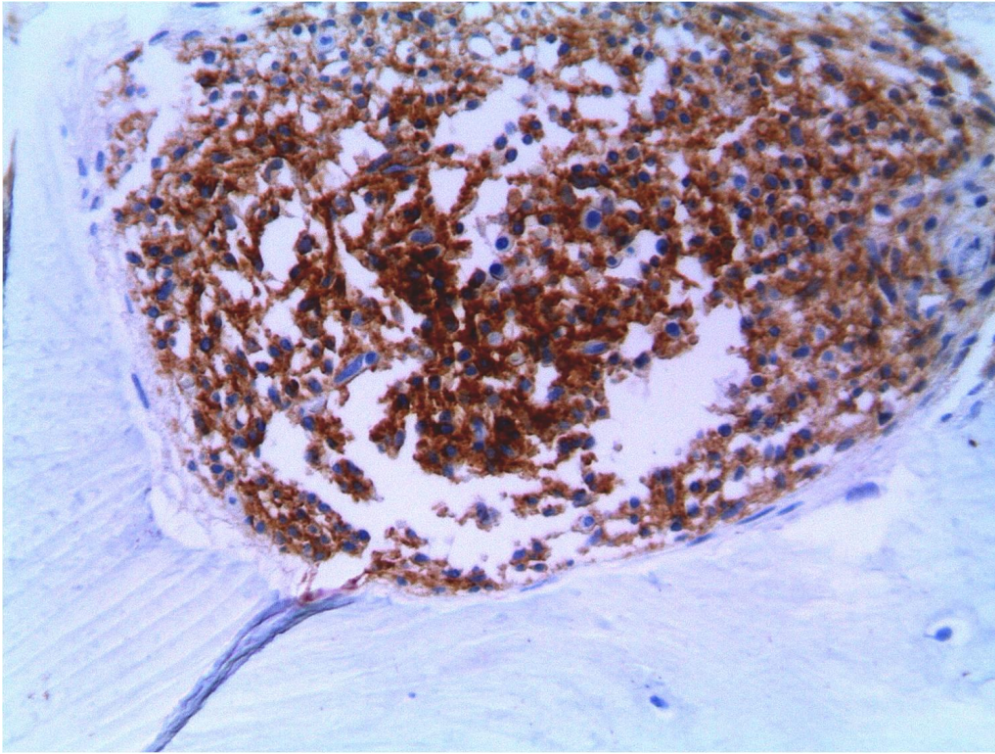
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Description: Sagittal CT scan MPR reformation showing diffuse vertebral sclerosis. **Origin:** Radiology Department, Hospital Povisa, Vigo, Spain

Figure 5

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Description: Immunohistochemistry study shows aggregates of spindle cells without atypia, being positive to c-kit, representing mast cells. **Origin:** Pathology Department, Hospital Povisa, Vigo, Spain