SYSTEMIC MASTOCYTOSIS AFFECTING THE SKELETAL SYSTEM

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Case report. A 56-year-old woman was being treated for dryness of her skin, pruritus and attacks of flushing. She was noted to have marked cyanosis of her hands and feet. Routine investigations revealed that she was in mild renal failure and an intravenous urogram showed multiple sclerotic bone lesions in the lumbar spine and right femoral neck (Figs 1 and 2). A skin biopsy stained with Giemsa, showed mast cells in excess of the normal number and histology of the femoral neck was also interpreted as being consistent with mastocytosis. Treatment with histamine blockers (cimetidine and cyproheptadine) was commenced. A two-month course has so far improved her pruritus but her unusual acrocyanosis remains.

Discussion. Systemic mastocytosis is a rare condition characterised by infiltration of various organs by mast cells. Benign and malignant forms are described, the benign existing in cutaneous and systemic varieties. The cutaneous form is mostly seen in childhood. The mast cell naevus is usually present at birth, while the more generalised urticaria pigmentosa is at about three to nine months. Both forms are usually self-limiting, but 10% of the latter may progress to systemic mastocytosis.

Adults usually present with cutaneous manifestations, but 50% progress to the systemic form of the disease which affects skin, bone, liver, spleen and the gastrointestinal tract. There is a 5% incidence of malignant transformation to mast cell leukaemia (Rafii et al. 1983), which responds poorly to treatment.

Mast cell proliferation particularly affects the reticulo-endothelial system. The cells secrete a variety of pharmacologically active agents, including histamine, heparin, prostaglandins, serotonin and mucopolysaccharides. These substances play an important role in the variety of presentations of the disease.

Skeletal abnormalities have been described in 10% of patients with systemic mastocytosis. Osteoporosis has been thought to be secondary to chronic heparin release and secretion of prostaglandin D2 by the mast cell. The latter may also be significant in the cause of acrocyanosis in the case presented (Kooette, Haak and Roberts 1983).

The histological diagnosis depends on the special metachromatic staining of the distinctive mast cell granules with Giemsa or toluidine blue. The biopsy specimens need careful handling to prevent degranulation. Because of the patchy nature of the disease and the difficulties with staining, positive marrow biopsies are often difficult to obtain and 24-hour urine collections for metabolites as well as isotope bone scanning are useful. Technetium and gallium scans demonstrate diffuse generalised uptake (Ensslen, Jackson and Reid 1983) and gallium is particularly taken up by the mast cell.

In conclusion, systemic mastocytosis should be considered in patients with painless sclerotic bone lesions or progressive osteoporosis. The importance of a skeletal survey and isotope bone scanning in patients with persistent urticarial skin conditions is stressed as is the careful handling of biopsy specimens.

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REFERENCES

