Case Report 61

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Fig. 1. An anteroposterior film of the lower extremities obtained at the age of 5 months demonstrates marked osteoporosis, characterized by considerable diminution in density of the bones and marked cortical thinning. Expansion of the medullary cavities with modeling abnormalities of the long bones is grossly apparent. The severe osteoporosis was also noted to involve most of the skeleton, with many compression fractures of thoracic and lumbar vertebral bodies.

Fig. 2. A follow-up anteroposterior roentgenogram of the lower extremities obtained approximately five years later shows almost complete remodeling of the tibiae and fibulae. The marked osteoporosis has regressed considerably, with increase in bony density of the shafts of the left tibia and fibula. Subcortical sclerosis and endosteal scalloping of the right tibia are noted. Small, well defined radiolucencies are observed in the ossification centers around the knees.

History

A 5-month-old Mexican boy presented with an urticarial-like rash involving the back, face and lower extremities. The rash was characterized by well-demarcated reddish patches, varying in size and shape. These patches faded within several hours after stroking of the skin, but would recur spontaneously.

Hepatosplenomegaly was present on physical examination. Hematological studies demonstrated the presence of anemia and thrombocytopenia.

A bone marrow aspiration was performed.

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Histological Sections

Fig. 3A and B. Low (A) and high power (B) photomicrographs of the histological sections of the bone marrow aspirate demonstrate multiple mast cells containing granular cytoplasm.

Diagnosis: Systemic Mastocytosis

The differential diagnosis must include Gaucher disease, Niemann-Pick disease, conceivably other storage disorders and thalassemia. Even osteogenesis imperfecta might also be considered in the initial films.

The bone marrow aspirate showed approximately 60 percent of the cells to be mast cells.

Discussion

Mastocytosis is an uncommon entity first described as a dermatologic disorder by Nettleship in 1969. Although originally classified as a skin disease it is now recognized as a systemic process with a wide spectrum of presentations. Multiple organ systems may be involved, including the skeleton, liver, spleen, lymph nodes and gastrointestinal tract. Cutaneous involvement is by far the most common manifestation. Approximately 75 percent of all cases of mastocytosis develop during infancy or childhood. Systemic involvement, however, is more likely to occur when the disorder first appears in adult life. The course of the disease is usually benign, but approximately one-third of the cases progress to what is termed mast cell leukemia.

The exact cause of the disorder is unknown, but the pathophysiology has been well documented and the systemic effects of this entity appear to be due to mast cell proliferation. The clinical features are correlated with histamine release which may cause local urticaria, flushing, diarrhea and vomiting. Peptic ulcer also may develop. The symptoms are not infrequently confused with those of the carcinoid syndrome. Hepatitis, splenomegaly and lymphadenopathy are commonly encountered. The hematological picture may vary, but anemia, leukopenia, leukocytosis and thrombocytopenia are reported. Hemorrhagic tendencies have also been noted in this entity.

The pathological findings deal with the effect of the proliferating mast cells. The pigmented, pruritic rash of urticaria pigmentosa is produced by the infiltration of the skin by the mast cells, which are of mesenchymal origin and are presumably basophilic cells. These cells are normally present in limited numbers in the connective tissues and the perivascular spaces. Mast cells elaborate histamine, serotonin, heparin and hyaluronic acid.

The mast cells have a significant effect on the skeleton, producing fibroblasts and granulomatous foci which replace bone marrow and destroy bony trabeculae. Osteoporosis and bone destruction result in the early stages, followed by generalized osteosclerosis.

Radiologically, localized and diffuse forms of skeletal lesions exist, which may be either lytic or blastic. It should be emphasized, however, that a mixed pat-