A 46-year-old man known to have diabetes insipidus had a two-day history of hemoptysis. Physical examination revealed a barrel chest with increased percussion note and widespread inspiratory crepitations at the bases. Sputum cytology was negative for malignancy, and there was no growth on culture. Two chest x-ray films are shown, one five years previously (Fig 1) and one on admission (Fig 2).
Diagnosis: Histiocytosis X

The two chest x-ray films show characteristic and progressive changes of interstitial pulmonary fibrosis. On both films however, there are also infiltrative changes in the inferior angles of the scapulae, with widening and deformity of most ribs. The most likely cause of these changes, together with the history of diabetes insipidus, is histiocytosis X, of which this patient has the Hand-Schuller-Christian variety. The skull (Fig 3) and left shoulder (Fig 4) also show the characteristic bone changes of mixed lysis and sclerosis which affected the entire skeleton apart from the hands and feet.

The underlying abnormality in histiocytosis X is granulomatous infiltration of large histiocytes with pale eosinophilic foamy cytoplasm. The Hand-Schuller-Christian variety usually begins before five years of age and is often first manifest by osteolytic bone lesions. Multisystem disease follows, with development of skin lesions, otitis media, adenopathy, splenomegaly, and lung infiltrates with fibrosis. The classic triad of skin lesions, exophthalmos, and diabetes insipidus may be encountered. In the lungs, granulomatous infiltrate of alveolar and bronchial walls with foamy histiocytes is found. This subsequently leads to fibrosis. As in other forms of lung fibrosis, there is an increased incidence of pneumothorax, hemoptysis, and "scar" carcinoma.

The diagnosis of the condition is by lung and bone biopsy. Treatment is with small doses of steroids and antineoplastic drugs such as methotrexate and vinblastine. In spite of widespread disease, patients may recover after many years, being left with residual orthopedic problems and diabetes insipidus.

References