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Pulmonary Gaucher's disease: high-resolution computed tomographic features

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Abstract. CT findings in pulmonary Gaucher's disease have not been previously reported. Chest radiograph of a patient with pulmonary involvement in type I Gaucher's disease proven by biopsy showed linear and reticulo-nodular opacities. High-resolution CT demonstrated thickening of the interlobular septa and between four and six small nodules within secondary lobules, probably each corresponding to an acinus.

Case report

A 21-year-old man was admitted with malaise and left flank pain of 7 months' duration. His physical examination revealed hepatosplenomegaly and the laboratory profile was normal except for a slight normochromic anemia. Type I Gaucher's disease was clinically suspected. Activity of the enzyme beta-glucocerebrosidase was decreased while serum angiotensin converting enzyme was increased to 488 U/l (almost ten times normal).

The admission chest radiograph showed diffuse and bilateral fine linear and reticulo-nodular densities (Fig. 1). High-resolution CT (HRCT) of the chest disclosed a combination of prominent septal lines and small nodular opacities throughout both lungs (Fig. 2). The patient had no respiratory symptoms. Pulmonary function tests were normal: FVC was 4.441 (96 % of predicted normal value) and FEV^1 was 3.691 (94 % of predicted normal value). Transbronchial lung biopsy demonstrated moderately thickened alveolar walls due to infiltration by lipid-laden macrophages and the alveolar spaces were tightly packed by the same cells (Fig. 3).

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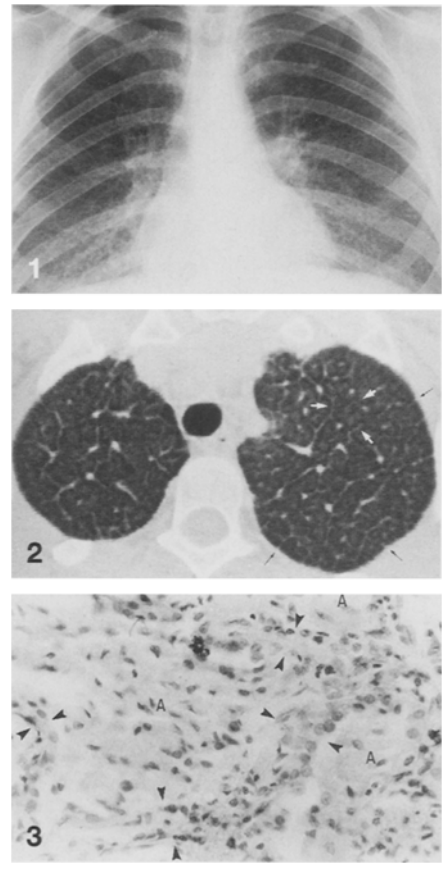
Fig. 1. Admission chest radiograph shows diffuse and bilateral fine linear and reticulo-nodular changes. The findings are seen predominantly at the lung bases

Fig. 2. High-resolution CT scan of the lungs. There is thickening of the interlobular septa peripherally *(black arrows)* and within the lung *(white arrows)*. There are small airspace nodules within several secondary lobules, each probably corresponding to an individual acinus

Fig. 3. Transbronchial biopsy specimen. The alveolar walls *(between arrowheads)* are thickened by infiltration of lipid-laden macrophages (Gaucher cells). Alveolar spaces *(A)* are packed with the same cells. (H & E × 310)



Discussion

Gaucher's disease is a genetically determined disorder of lipid metabolism. The basic defect is deficiency of the enzyme beta-glucocerebrosidase, as a result of which glucocerebroside accumulates within the reticuloendothelial cells, histiocytes and macrophages (Gaucher cells). The liver, spleen, bone marrow, brain or lungs may be infiltrated by Gaucher cells.

Depending on the age of onset and presence or absence of central nervous system involvement, Gaucher's disease is classified into three distinct forms [1, 2]. In the adult form (type I) the central nervous system is intact and first manifestations of the disease (commonly hepatosplenomegaly) may appear at any age. The course is relatively benign. The infantile form (type II) is characterized by early (typically before 6 months of age) central nervous system involvement and death within 2 years. The juvenile form (type III) is a subacute variant of the disease which comprises cases with combined involvement of the central nervous system and the other organs. Pulmonary involvement is not unusual

in the infantile form, but is particularly rare in the adult form [2].

In the lung, lipid-laden macrophages (Gaucher cells) aggregate within the alveolar spaces and infiltrate alveolar walls, perivascular and peribronchial spaces, and pulmonary septa [2–4]. Occasionally the precapillaries and capillaries may be occluded by Gaucher cells, leading to perfusion defects and pulmonary hypertension [1].

At the early stage of lung involvement, the pulmonary function tests remain normal although impressive histologic changes may exist. However, pulmonary disease is progressive and patients usually succumb to the disease with infection superimposed on respiratory failure. Pulmonary hypertension may be an additional factor in the patient's rapid demise.

Plain chest radiographic findings in pulmonary Gaucher's disease are described as miliary [1], reticular [2], nodular [1, 3] or reticulo-nodular [1, 5, 6] opacities. In advanced cases the infiltrates conglomerate and form larger densities [3].

HRCT features of our patient correlated well with the underlying pathology. Thickening of the septa reflect infiltration of the pulmonary interstitium by Gaucher cells. Clusters of four to six small nodular opacities surrounded by interlobular septa represent accumulation of the same cells within airspaces, each nodule most probably corresponding to an acinus. Thus, pulmonary Gaucher's disease enters into the differential diagnosis of combined interstitial and airspace opacities seen on HRCT studies of the chest.

To the best of our knowledge this is the first CT description of pulmonary complications of Gaucher's disease. We believe, because of the recent development of hepatosplenomegaly, the absence of respiratory symptoms and the preservation of the pulmonary architecture, CT findings described in this case represent early manifestations of pulmonary involvement. Hopefully, the spectrum of CT features of this rare disease will be defined by further case reports.

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Continued from p. 234

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