Radiological Findings of Pulmonary Involvement of Type B Niemann-Pick Disease

Hallazgos radiológicos de afectación pulmonar por enfermedad de Niemann-Pick tipo B

To the Editor:

Niemann-Pick disease is a recessive, autosomal hereditary lysosomal storage disease. Deficient activity of acid sphingomyelinase causes build-up, mainly sphingomyelin, in central nervous system and reticuloendothelial system tissues. Depending on which organs are affected, there are various clinical subdivisions of Niemann-Pick disease. The most severe of the six disease subtypes is type A, which has an acute neuronopathic phenotype tending to cause death in early childhood. Type B is a heterogeneous disorder that presents in most patients without affecting the nervous system. Progressive pulmonary disease, hepatosplenomegaly, short stature and pancytopenia may be present where involvement is systemic.

Our patient is a seven-year old girl. At nine months old, she was examined in the hospital due to abdominal distension. Hepatosplenomegaly was observed and the patient underwent a fine-needle aspiration biopsy of the liver. The diagnosis was Niemann-Pick type B. She presented dry cough and a fever. Physical exploration revealed hepatomegaly and splenomegaly. Cardiovascular and neurological examinations were normal. Pulmonary auscultation revealed pronounced crackling stertor in the lung bases. The kidney and liver function tests were normal. Laboratory findings revealed mild anaemia. After a clinical examination, the patient was referred to our centre for a chest radiography and high-resolution computed tomography (HRCT). Postero-anterior radiography revealed diffuse reticulonodular infiltration at the base of both lungs. The HRCT tests were performed without intravenous contrast. A multi-slice spiral computed tomography scan was performed using a 16-slice CT system (Somatom Sensation 16, Siemens, AG, Erlanger, Germany). Gantry rotation time was 0.5 s. We used a tube with 100kVp voltage and a tube with 40mA (effective) current. The slice thickness was 1 mm, with a slice interval of 9 mm. An edge-detection filter was used.

EBUS easily detects mediastinal cysts: it distinguishes them from vascular structures due to their lack of Doppler flow signal, and also enables cytological diagnosis and treatment by draining.

In our case, we observed a ground-glass appearance, with lipid storage disorders, especially in children. The most reliable radiological technique to confirm it is HRCT (Fig. 1).

In conclusion, we must consider the possibility of a lung condition caused by a mediastinal cyst. Although this is not the most common profile, Niemann-Pick type B should be added to the list of diseases that can present this appearance. Progressive pulmonary infiltration is an important cause of morbidity and mortality. To date, no successful treatments for lung damage caused by Niemann-Pick disease have been recorded. Complete pulmonary lavage seems to be a potentially useful treatment.

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References


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Figure 1. a) The postero-anterior radiography shows diffuse reticulonodular infiltrate in the base of both lungs. b) Close-up of reticulonodular infiltrate affecting the lower parts of the lung. c) High-resolution computed tomography (HRCT) showing a ground-glass section accompanied by septal interlobular-intralobular thickening and peribronchovascular interstitial thickening. d) HRCT of the superior areas of the lung showing subpleural nodules.