Pediatr Radiol (1992) 22: 78–79

Pediatric Radiology

Gaucher's disease

Plain radiography, US, CT and MR diagnosis of lungs, bone and liver lesions

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Received: 23 August 1991; accepted: 20 September 1991

Abstract. We report our observations made by conventional radiography, ultrasound, computerized tomography (CT), and magnetic resonance imaging (MRI) on a 3½-year-old girl with Gaucher's disease. The interest of the case consists in the exceptional lungs involvement, the demonstration by MRI of the bone marrow involvement and the necrosis and fibrosis of the liver, as shown by CT. This liver complication has been previously reported only once.

Gaucher's disease is an hereditary lysosomal storage disease in which a deficiency of the enzyme glucocerebrosidase is associated with accumulation of the enzyme substrate, glucosylceramide, in reticuloendothelial cells throughout the body, primarily in the liver, spleen, lymph nodes and bone marrow. The diagnosis rests on the demonstration of the pathognomonic Gaucher cells in the infiltrated organs and is confirmed by assaying glucocerebrosidase activity in white cells or fibroblasts. There are 3 clinical variants of Gaucher's disease corresponding to genetic variants of the enzyme defect [1]. We wish to report a particularly aggressive form of type I (chronic nonneuronopathic) Gaucher's disease with exceptional lungs and liver involvement.

Case report

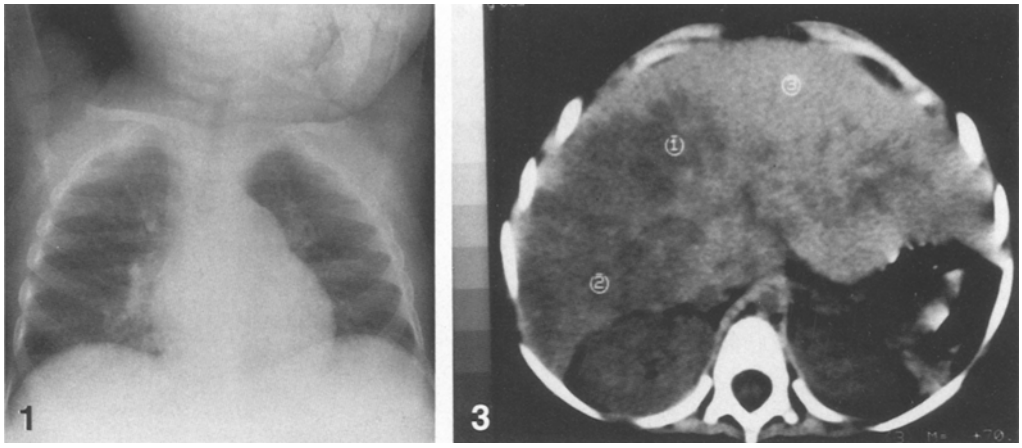
A 3½-year-old turkish girl was hospitalised for the first time at the age of two presenting with severe pallor, poly adenopathy and enlargment of the liver and spleen. The neurological examination was normal. Blood analyses revealed pancytopenia and a slight increase in transaminases. A bone marrow and a lymph node biopsy both demonstrated the presence of gaucher cells. The diagnosis is confirmed by the low level of glucocerebrosidase enzyme activity in white blood cells and fibroblasts. In the following months thrombocytopenia and the extent of the splenomegaly necessitated a splenectomy. A liver biopsy taken at the time of surgery showed diffuse hepatic fibrosis. A year later, the child was referred for bone marrow transplant. The clinical state of the child deteriorated, the bone marrow transplant was rejected and the child died of liver and heart failure.

Radiographic features

Plain radiography of the chest revealed a reticulonodular pattern affecting both lungs diffusely (Fig. 1). Bone marrow infiltration by Gaucher cells is associated with osteopenia, cortical thinning and osteolytic lesions which were particularly visible in this case in the humeral diaphyses. The lower extremities of both femoral shafts had the characteristic "Erlenmeyer flask" deformity (Fig. 2a). MR imaging exam of the knee was performed: both the T1 weighted image (Fig. 2b) and the T2 weighted FFE image (Fig. 2c) demonstrated an abnormally low signal intensity for the bone marrow as well as a zone of pathologically high signal intensity on the T2 weighted FFE image. Ultrasound and CT of the liver showed huge hepatomegaly and adenopathy at the level of the hilum. On ultrasound, the liver was homogenous but with decreased echogenicity. Unenhanced CT showed inhomogeneous liver density with hypodense zones within the right hepatic lobe (Fig. 3).

Discussion

Chest radiograph is generally normal in Gaucher's disease. In a review of the literature in 1975, Wolson found only 10 cases with pulmonary involvement: a bilateral diffuse reticulonodular or miliary pattern of pulmonary infiltrate is described as typical radiographic appearance [2]. Since then about ten additional cases of lung involvement have been reported. The radiologic literature has stressed the classic bone changes [3]. The great sensitivity of MRI allows the detection of abnormalities of the marrow in patients with Gaucher disease even when plain radiographs or CT scans showed no abnormality. In children, knowledge of the normal pattern of red to yellow conversion with age is essential before pathologic processes can be identified. Moore and Dawson reported the spectrum of MR appearance of femoral marrow with age on T1 weighted images in children. Between the ages of 1 and 5 years, histologically, the diaphyseal marrow still maintains a large proportion of red marrow, but the increased signal intensity from fatty marrow appears to predominate to give an overall MR appearance of yellow marrow. This is a reflection of the sensitivity of T1 weighted images to microscopic fat present in marrow that appears red macroscopically [4]. Infiltration of the bone marrow by Gaucher cells results in an abnormally low signal intensity on both T1 and T2 weighted images indicating a hypercellular marrow. The epiphyses are generally spared unless the involvement of bone is extensive [5]. In our observation, MR of the knees shows abnormal low signal intensity, with involvement of the epiphyses. On T2 weighted FFE image, marrow edema is seen in the center of the diaphysis as an increase in marrow signal intensity. On ultrasound, generally no abnormality is noted in the liver other than diffuse hepatic enlargement [6]. In our case, there was no focal lesion in the



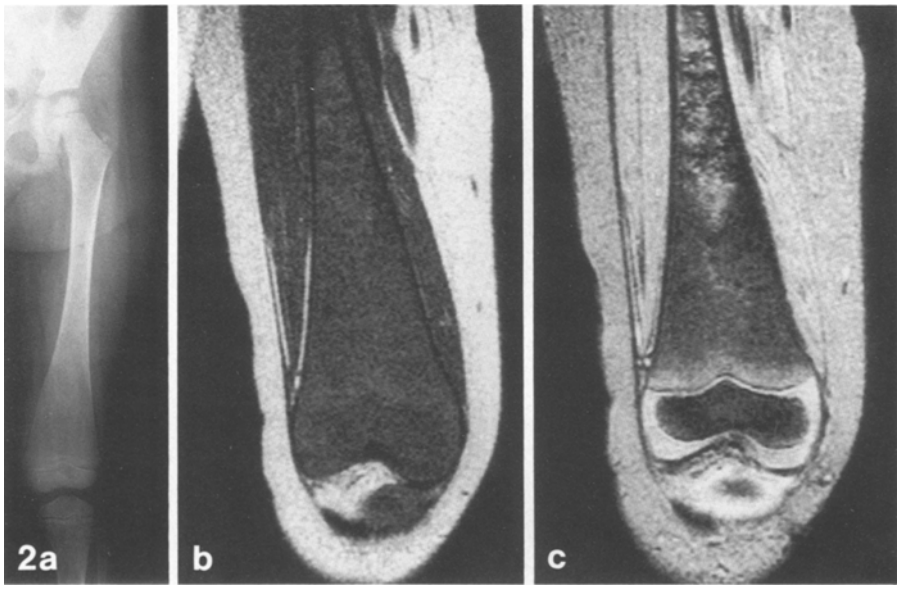


Fig. 1. Chest X-ray reveals bilateral diffuse reticulonodular infiltrates

Fig. 2. **a** Plain radiograph shows characteristic "Erlenmeyer flask" deformity of the lower extremity of the femoral shaft. **b** T1 weighted (650/25) coronal MR image through the femur demonstrates an abnormally low signal intensity for the bone marrow, including the distal epiphysis. **c** T2 weighted FFE (475/14) coronal MR image through the femur. Marrow edema is seen as an increase in marrow signal intensity

Fig. 3. Unenhanced CT of the liver shows huge hepatomegaly and hypodense zones in the right hepatic lobe

liver on US examination although CT scan showed inhomogeneous liver den sity with hypodense zones in the right lobe. Liver biobsy and the CT appearance suggests that these regions are areas of necrosis and fibrosis. Only one similar case has been reported before [7]. In conclusion, various types of radiological imagery have allowed us to establish the extent of infiltration of several organs in a particularly aggressive form of Gaucher's disease. We report findings that are rarely described of involvement of the lungs and liver which suggested a most unfavourable prognosis. We also highlight the advantages in using MRI to evaluate infiltration of the bone marrow by pathologic cells.

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