

Liver scintigraphy in a patient with Gaucher disease

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Liver scintigraphy including SPECT was performed in a patient with Gaucher disease and compared with other methods. Multiple photon-deficient areas in the liver and spleen were recognized, and in evaluating the reticuloendothelial system of the liver, liver SPECT image was superior to US, CT and MR images.

Key words: Gaucher disease, liver, SPECT, MRI

INTRODUCTION

GAUCHER DISEASE (Glucosyl cerebroside lipidosis) is a familial disorder of lipid metabolism due to a lack of glucocerebrosidase activity resulting in an accumulation of glucocerebrosides in reticuloendothelial cells. It is the most frequently seen lipidosis but uncommon, and clinically it is manifested by hepatosplenomegaly, skin pigmentation, skeletal lesions and pinguiculae.¹

In this paper, a case of Gaucher disease is described with liver scintigraphy including SPECT, US, CT and MR.

CASE REPORT

A 30-year-old male was referred to our hospital due to 8 months history of right hip joint pain. His elder brother was diagnosed as having Gaucher disease. Surgery for aseptic necrosis of the right femoral head was scheduled. Preoperative examination revealed thrombocytopenia ($4.7 \times 10^4/\text{mm}^3$). Other laboratory data are shown in the table. Ultrasonography (US) (Aloka SSD-650) revealed marked splenomegaly with multiple hypoechoic areas (Fig. 1) and hepato-

megaly with no definite focal changes. Contrast-enhanced CT (Yokogawa CT 9000) demonstrated multiple low density areas in the enlarged spleen, but no focal changes in the enlarged liver (Fig. 2).

In MR [Toshiba MRT 2000 FXII (1.5T)] examination, both T₁-weighted (600/13) and T₂-weighted (2,500/80) images demonstrated multiple low intensity areas in the spleen. Low intensity areas in the liver were also suspected, but were not so clear (Fig. 3).

Subsequently performed bone-marrow aspiration revealed Gaucher cells (Fig. 4) and the glucocerebrosidase activity of neutrophil was low. The diagnosis of Gaucher disease was therefore confirmed. To evaluate the reticuloendothelial system of the liver and spleen, liver scintigraphy with 111 MBq ^{99m}Tc-phytate was performed and showed hepatomegaly and marked splenomegaly with heterogeneous tracer distribution (Fig. 5A). SPECT (Toshiba GCA-901A) was then performed. 60 views were obtained at 6° angular increases with an acquisition time of 10 seconds per view. Before reconstruction, the initial planar images were prefiltered with a Butterworth filter (frequent 0.11, order 8). Filtered backprojection was performed with a Ramp filter. A Chang attenuation correction was performed. Multiple photon-deficient areas in the enlarged liver and spleen were more clearly demonstrated (Fig. 5B).

Splenectomy for thrombocytopenia and wedge biopsy of the liver was performed. The weight of the spleen was 2,500 g. Infiltration of Gaucher cells to

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Fig. 1 Ultrasonogram revealed scattered hypoechoic lesions in the enlarged spleen (5 MHz).

Table 1 Preoperative laboratory data

RBC	$403 \times 10^4/\text{mm}^3$	
WBC	$4,100/\text{mm}^3$	
Hb	14.2 g/dl	
Ht	40%	
Plt	$4.7 \times 10^4/\text{mm}^3$	↓
TP	7.9 g/dl	
Alb	4.1 g/dl	
GOT	30 IU/l	
GPT	18 IU/l	
Al-P	156 IU/l	
LAP	48 IU/l	
T Bill	1.5 mg/dl	
D Bil	0.5 mg/dl	
TTT	7.8 MU	↑
ZTT	17.5 KU	↑↑
ChE	0.43 Δ PH	↓
BUN	13.6 mg/dl	
Cr	0.8 mg/dl	

the liver and spleen was revealed histopathologically (Fig. 6).

DISCUSSION

Accompanying the development of US, CT and MR, liver scintigraphy seems to decrease in clinical usefulness as a means of morphological examination due to its poor space resolution.

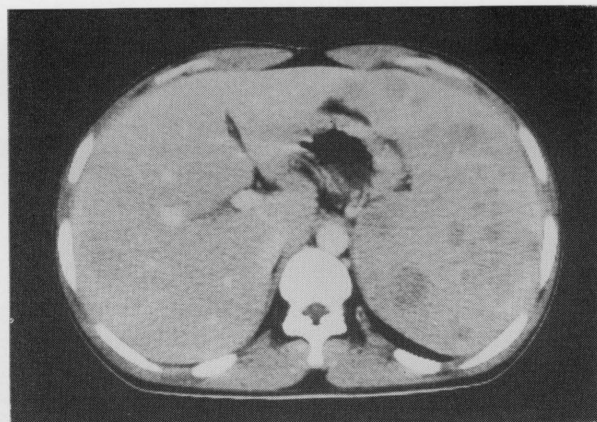


Fig. 2 Contrast-enhanced CT demonstrated multiple low density areas in the enlarged spleen, but no focal changes in the liver were shown.

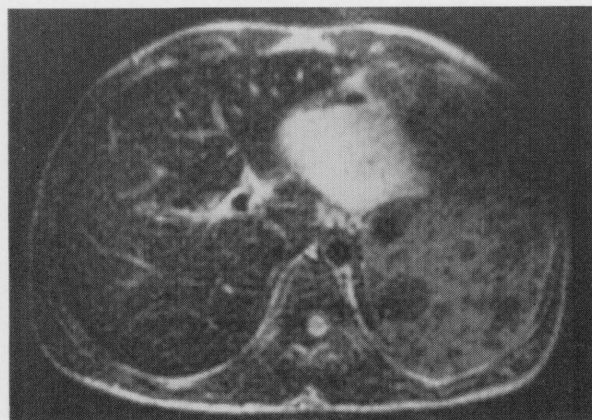


Fig. 3 T₂-weighted (2,500/80) MR images demonstrated multiple low intensity areas in the spleen, but liver lesions were not clear.

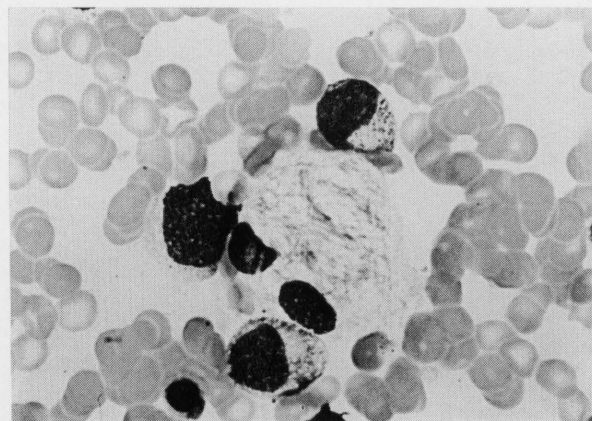
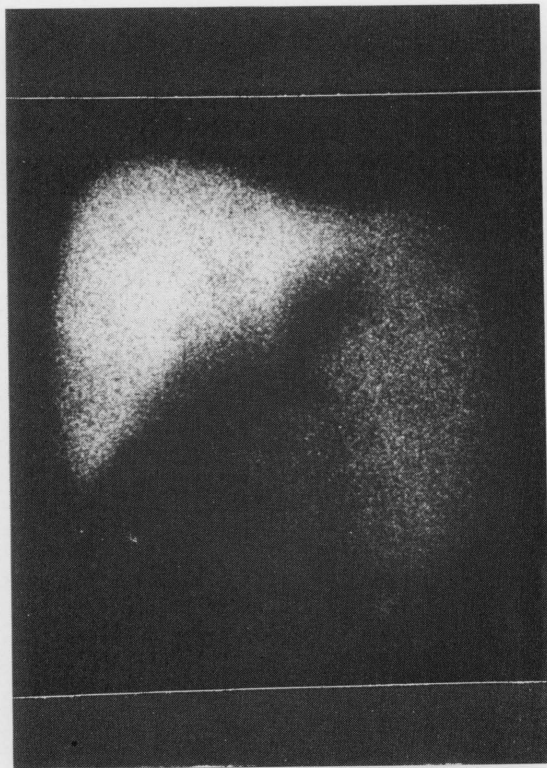


Fig. 4 Bone-marrow aspiration revealed Gaucher cell.

Hill, et al. reported US findings in the spleen of 52 patients with Gaucher disease revealing that most patients had discrete hypoechoic lesions that corresponded pathologically to clusters of Gaucher cells.² Aspestrand, et al. reported focal changes in the spleen of a case of Gaucher disease. Multiple low-



A



B

Fig. 5 A, Planar liver scintigraphy showed marked hepatosplenomegaly with heterogeneous tracer distribution. B, SPECT showed multiple photon-deficient areas in the liver and spleen.

attenuating lesions in the enlarged spleen were shown by contrast-enhanced CT and numerous contrast defects in the enlarged spleen were shown by celiac angiogram, and these findings were reported to correspond to the reduced vascularity of the lesions.³ Rosenthal, et al. reported the MR finding of skeletal involvement of Gaucher disease.

MR spectroscopy performed on fresh tissue immediately after operation has revealed significantly lower T_2 values in patients with Gaucher disease than

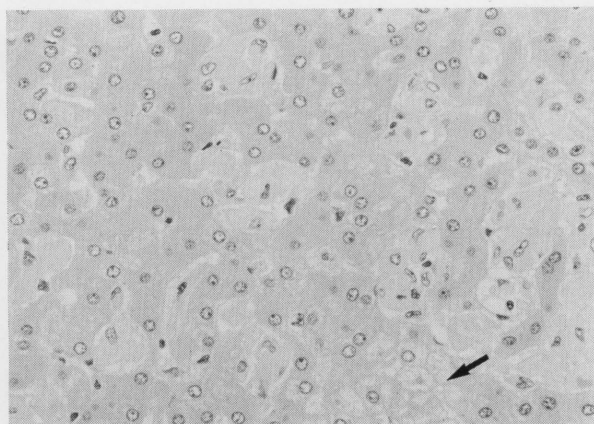


Fig. 6 Infiltration of Gaucher cells to the liver was recognized (arrow).

in normal control patients.⁴ And this reduced T_2 value is responsible for the characteristic low signal intensity.^{4,5} MR imaging enhanced with superparamagnetic iron oxide, which is applied as a reticuloendothelial contrast agent, may be a promising method,⁶ but we have not been able to use this contrast agent yet. About scintigraphic findings in Gaucher disease, it has already been reported that liver scintigraphy shows various degrees of enlargement and inhomogeneous tracer accumulation in the liver and spleen, and seems to be a simple and sensitive test for evaluating the liver and spleen. However, only planar images of the liver and spleen have been reported.^{7,8} In the present study, splenic lesions were well detected by US, CT, MR and SPECT. However, in evaluating liver lesions, SPECT was superior to US, CT and MR.

The reason why US and CT could not detect liver lesions but could detect splenic lesions was not clear. Since liver lesions of Gaucher disease are caused by an accumulation of glucocerebrosides in reticuloendothelial cells, liver scintigraphy, which reflects phagocytic activity of Kupffer cells, is thought to be useful. Unlike liver cirrhosis, no bone-marrow activity is observed in Gaucher disease, which indicates that the liver has sufficient reticuloendothelial reserve and the marrow is infiltrated by Gaucher cells.⁸ In conclusion, spleen and liver lesions in a case of Gaucher disease were evaluated by means of US, CT, MR and scintigraphy including SPECT images. Splenic lesions were clearly detected by all methods, but in evaluating the reticuloendothelial system of the liver, SPECT was superior to US, CT and MR.

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