STUDY OF α-D-GLUCOSIDASE ACTIVITY IN PATIENTS WITH POMPE'S DISEASE

CHING-YUANG LIN, BETAU HWANG, KWANG-JEN HSIAO
and YI-RU JIN

龐佩氏病患者 α-D-Glucosidase 酶活性之研究

林清淵　黃碧桃　蕭廣仁．金一如

Reprinted from the Journal of the Formosan Medical Association
Vol 85, No 8, pp 766～770, August, 1986

臺灣醫學會雜誌第85卷第8號第766～770頁抽印本
中華民國75年8月發行

J Formosan Med Assoc 85; 766-770, 1986

STUDY OF α-D-GLUCOSIDASE ACTIVITY IN PATIENTS WITH POMPE'S DISEASE

CHING-YUANG LIN, BETAU HWANG, KWANG-JEN HSIAO and YI-RU JIN

Activities of 2 major forms of α-glucosidase in muscle cells, lymphocytes and cultured fibroblasts of patients with Pompe's disease and normal healthy controls were measured by using 4-methylumbelliferyl-α-D-glucoside as the substrate. The lowest enzyme activity was in muscle cells and the highest in cultured fibroblast. However, even in the muscle cells, there was no overlap between patients with Pompe's disease and the controls. These results indicate that enzyme determination from patient's muscle cells, lymphocytes, cultured fibroblasts is effective in the diagnosis and genetic counselling for Pompe's disease.

Key words: ***Pompe's disease, α-glucosidase.***

(*J Formosan Med Assoc* **85**; 766-770, 1986)

Glycogen storage disease type II (Pompe's disease) (Mckusick No. 23230) is an autosomal recessive disorder, characterized by the lysosomal accumulation of glycogen.[1,2] The impaired glycogen degradation is due to a deficiency of acid α-glucosidase (EC 3. 2. 1. 20).[1~8] Several clinical forms of Pompe's disease which differ in age of onset, organ involvement, and progression of the disease have been recognized.[2] In Taiwan, most of the cases were infantile form (generalized glycogenosis II), nearly all tissues were affected, and symptoms became apparent shortly after birth. Hepatomegaly and muscular weakness were present, and cardiac failure caused by the extensive accumulation of glycogen usually resulted in death within the first year of life.

In Taiwan, Pompe's disease is the most common type of glycogen storage disease. It is rare in Japan, Norway, Sweden, West Germany and the Netherlands.[9] It involves not only Chinese but also is found in a group of aborigines in Taiwan. In an attempt to gain the acid α-D-glucosidase activity in Chinese patients with Pompe's disease, we measured acid α-D-glucosidase activity in lymphocytes and cultured fibroblasts from these patients.

MATERIALS AND METHODS

Patient population

From April 1981 to August 1985, 13 babies suffering from Pompe's disease were studied. Their ages ranged from 50-days-old to 11 months old; 8 were males and 5 were females. The criteria for diagnosis of Pompe's disease were: (1) presence of clinical features of hypotonia, cardiomegaly and hepatomegaly; (2) the electrocardiogram showed depressed ST segments, inverted T waves and a shortened P-R interval; (3) muscle biopsy showed vacuolation on the glycogen-containing area in light microscopic examination and multigranular deposits of glycogen in the sarcoplasm and bound

Departments of Medical Research and Pediatrics, Veterans General Hospital, Taipei, Taiwan, Republic of China.
Received April 17, 1986.
Accepted June 12, 1986.