F-17 Proc. of 1st Pan-Pacific Congress of the Screening of Inborn Errors of Metabolism, Kanazawa, F-17, (1985)

POMPE'S DISEASE IN CHINESE AND THE PRENATAL DIAGNOSIS BY DETERMINATION OF α-GLUCOSIDASE ACTIVITY

Ching-Yuang Lin, Betau Hwang* and Kwang-Jen Hsiao

Pediatric Research Laboratory, Department of Medical Research and *Department of Pediatrics, Veterans General Hospital, Taipei, Republic of China

In Taiwan, Pompe's disease is the most common glycogen storage disease. The defective enzyme is α-D-glucosidase. The hereditary pattern is autosomal recessive trait.

During the 3 years period from 1983 to 1985, 12 cases of Pompe's disease (type II glycogenosis) were diagnosed by enzymatic, histological and ultrastructural studies. α-Glucosidase activity was assayed in lymphocytes from patient's peripheral blood, muscle cells and skin fibroblast with 4-methylumbelliferyl-α-D-glucopyranoside as the substrate. The acid α-glucosidase activity (pH 4) of all patients was less than 1 nmole/mg protein/hour. The pH vs activity curve of the muscle α-glucosidase in all patients showed a pattern similar to that of classical Pompe's disease reported in Caucasian patients. The histologic and ultrastructural picture of the muscle fiber in all cases revealed evidence of increased glycogen storage. Three of 12 mothers who have born a Pompe's disease child received amniocentesis in subsequent pregnancy. One of the fetuses was homozygote. The α-glucosidase activity in subculture amniotic cells was 0.73 nmole/mg protein/hour (pH 4). One was heterozygote and another one was normal. All of the prenatal diagnosis were confirmed by determination of

the acid α-glucosidase activity in the skin fibroblast culture taken after each birth.

These results indicate that determination of α-glucosidase could be used effectively for diagnosis and prevention of Pompe's disease.