Proceedings of First Conference of Asian and Oceanian Association of Child Neurology, Taipei, p.44, 1983

## F-33 LABORATORY TESTS FOR SCREENING, DIAGNOSIS AND MANAGEMENT OF PHENYLKETONURIA

*Kwang-Jen Hsiao[1,2], Nai-Yeh Hsu[3], Ching-Shiang Chi[3], Kuang-Dong Wuu[4] & Wen-Kuo Ting[2]

*Clin. Biochem. Res. Laboratory[1] and Depts. of Clin. Laboratory[2] & Pediatrics[3], Veterans General Hospital; Genetics Laboratory[4], National Yang-Ming Medical College; Taipei, Taiwan, R.O.C.*

It was reported that early diagnosis with proper dietary therapy could prevent the mental retardation caused by phenylketonuria (PKU). By using the Guthrie Bacteria Inhibition Assay (BIA) of blood phenylalanine (Phe) as the screening method, a 20 months and a 12 years old PKU boy were detected in Taiwan from 70 hospital out-patients and 551 children in mental institutions, respectively. Both of them were confirmed by urinary $FeCl_3$ & Phenistix tests, serum Phe (24.7—29.9mg/dl) & tyrosine (Tyr, 0.6—1.1 mg/dl) determined by fluorometric method, and plasma Phe/Tyr ratio (>31.6) determined by amino acid analyzer. Both of them were ruled out of biopterins metabolism defect by determination of 24 hr. urinary catecholamines (by HPLC). The $FeCl_3$ & Phenistix tests became negative when blood Phe were reduced to 15 mg/dl, which indicated that these tests were not sensitive enough for diagnosis nor for monitoring of therapy. The blood and serum Phe of three PKU patients on dietary therapy has been monitored by BIA and fluorometric methods, respectively. The fluorometric method was more sensitive and reproducible than the semi-quantitative BIA method. With these laboratory tests, we should be able to screen, diagnose and monitor the dietary therapy of PKU in Chinese children.