F-5

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A PILOT PROGRAM FOR AN INTERGRATED NATIONWIDE SYSTEM FOR NEONATAL SCREENING OF CONGENITAL METABOLIC DISEASES IN TAIWAN

Kwang-Jen Hsiao and Kuang-Dong Wuu*

Clinical Biochemistry Research Laboratory, Department of Medical Research, Veterans General Hospital, Taipei, Taiwan, China, *Genetics Laboratory, National Yang-Ming Medical College, Taipei, Taiwan, China

A pilot neonatal screening project has been carried out for developing a nationwide neonatal screening program for early diagnosis and treatment of congenital metabolic diseases in Taiwan. From 1984. 1. 1. to 1985 7.30, the capillary blood of 18,316 newborns was collected on filter paper about five days after birth by 13 hospitals form different parts of the island, Nan-Tou County health stations, and obstetric clinics and mid-wives in the northern districts of Hsin-Chu County. This collecting system represented all types of newborn delivery on this island. The effective collection rate was 95%. The dry blood samples were mailed to the screening center (Clin. Biochem. Res. Lab., VGH). Congenital Hypothyroidism (CHT), phenylketonuria (PKU), galactosemia, maple syrup urine disease and homocystinuria were screened by determination of TSH (EIA), phenylalanine (Guthrie), galactose (E.Coli-Phage) and transferase (Beutler), leucine (Guthrie), and methionine (Guthrie), respectively. Abnormal results were found in 647 specimens. The cases with high positive values were referred to one of the 5 local diagnosis and treatment centers immediately by phone calls. A second sample was requested in other positive cases and was collected by the sample collecting system or the follow-up system, which consists of public health nurses in every county on this island. If the result was still

positive, the case was then referred to local centers. Eight cases of CHT (one of them was transient), one case of PKU and one case of galactosemia were confirmed. All cases of CHT were diagnosed and treated within 23 days of life. The case of PKU, which is caused by biopterin synthesis deficiency, was diagnosed and treatment started at the 37th day of life. The lactose-free diet was started for the galactosemic case 10 days after birth. All of the cases are developing normally at the present time. These results show that this pilot screening program could provide very effective preventive medical service in Taiwan. With the high prevalence ($\sim$1/2500) of CHT in Taiwan, a nationwide neonatal screening program based on this model system to prevent those affected babies from mental retardation is highly indicated. (This study was supported by a grant from the Dept. of Health, Executive Yuan, ROC)