Proc Symposium on Molecular Approaches to Genetic Disorders, Taipei, 1989;P.16-18.

NEONATAL SCREENING FOR INBORN METABOLIC DISEASES IN TAIWAN

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In order to study the morbidity of potentially treatable congenital metabolic diseases in mentally retarded children and to establish screening tests, diagnostic procedures, and therapeutic methods, the blood of 551 institutionalized children and 4,744 mentally retarded school-children in Taiwan were collected on filter paper between 1982 and 1983. Congenital hypothyroidism (CHT), phenylketonuria (PKU), galactosemia, maple syrup urine disease (MSUD), and homocystinuria were screened by determination of TSH (EIA), phenylalanine (Guthrie), galactose (E. Coli-Phage) and transferase (Beutler), leucine (Guthrie), and methionine (Guthrie) in the dried blood spots, respectively. Four cases of PKU and 10 cases of CHT were detected. The morbidity of PKU and CHT in mentally retarded Chinese children was estimated at around 0.04-0.2% and 0.15-0.5%, respectively. Therefore, A pilot project has been carried out for developing a nationwide neonatal screening program for early diagnosis and treatment. From 1984.1 to 1985.7 the capillary blood of 18,316 newborns was collected on filter paper about five days after birth by 13 hospitals, Nan-Tou County health stations, and

(107)

obstetric clinics and mid-wives in 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 and the five congenital metabolic diseases were screened. The cases with high positive values were referred to one of the 4 local diagnosis and treatment centers immediately by phone calls. A second sample was requested in borderline 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 PKU and one glactosemia were confirmed. All cases of CHT and galactosemia 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. All of the cases are developing normally at the present time.

The integrated neonatal screening system developed by the pilot project was adopted to be the nationwide service program by the Dept. of Health in lated 1984. Two screening centers was established and more than 50% of newborns (approx. 150,000) were screened in the 1988 fiscal year. From 1984.1 to 1988.11, 194,163 newborn samples collected from ~~270~~ 249 delivery units were screened by our center. One hundred and eighteen CHT (1/2,000),

5 PKU (1/40,000), 1 galactosemia and 1 homocystinuria were detected. Most of them were diagnosed and treated within 3 weeks of life. For studying the screening of glucose-6-phosphate dehydrogenase (G6PD) deficiency, a bolld spot semi-quantitative flourometric method was developed to back up the qualitative spot test. The incidence of G6PD deficiency is estimated to be around 2% in Taiwan from 186,754 newborns screened. Eleven local G6PD referral centers connected with fax were established for following-up positive cases. Since no MSUD was found from 200,000 neonates screened in Taiwan. After two-year (1985.7-1987.6) pilot study on G6PD screening, the MSUD was replaced by G6PD in the routine neonatal screening program.