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SY35 NEONATAL SCREENING FOR INBORN METABOLIC DISEASES: THE EXPERIENCE 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, between 1982 and 1983 blood from 551 institutionalized children and 4,744 mentally retarded school-children in Taiwan was collected on filter paper. 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), 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 was carried out with a view to develop a nationwide neonatal screening program. From January 1984 to July 1985 capillary blood from 18,316 newborns was collected on filter paper about five days after birth in 13 hospitals, Nan-Tou County health stations, and obstetric clinics and by 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 for screening for the five congenital metabolic diseases. The cases with high positive values were referred to one of the 5 local centers immediately by phone. 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 the local center. Eight cases of CHT, one of PKU and one of 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 the cases are developing normally at the present time.

The integrated neonatal screening system developed by the pilot project was adopted as the nationwide service program by the Dept. of Health in late 1984. Two screening centers were established and more than 50% of newborns (approx. 150,000) were screened in the 1987 fiscal year. From January 1984 to December 1987, 123,227 blood samples collected from 152 delivery units were screened by our center. Sixty-two CHT (1/2,000), 3 PKU (1/42,000), 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 blood 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 115,818 newborns screened. Eleven local G6PD referral centers connected by fax were established for the follow-up of positive cases. After a two-year (July 1985-June 1987) pilot study on G6PD screening, the MSUD was replaced by G6PD in the routine neonatal screening program, since no MSUD was found in 200,000 neonates screened in Taiwan.

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