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NEONATAL SCREENING OF PHENYLKETONURIA IN TAIWAN

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A nationwide program for neonatal screening of congenital metabolic diseases in Taiwan was started in 1984. There were two screening centers, one located at Veterans General Hospital-Taipei (VGH) and the other located at National Taiwan University Hospital. In 1992, 94% (302,571) newborns delivered in Taiwan were screened by these two centers. From 1984.1 to 1993.1, 838,565 newborns were screened for phenylketonuria (PKU) by the VGH screening center. Dried blood spot samples collected on filter paper by heel puncture were sent to our screening center from 445 delivery units, including 111 hospitals, 260 obstetric clinics, 23 midwives and 51 health stations, all over Taiwan by mail. Phenylalanine levels in the dried blood samples were determined by the bacterial inhibition assay (Guthrie test). The case with high positive values (Phe > 4 mg/dl) were recalled immediately by phone calls. A second sample was requested in borderline positive (Phe 2-4 mg/dl) cases. The recalled cases were differentially diagnosed by measurement of blood Phe and Tyr concentrations, analysis of urinary organic acids, determination of blood dihydropteridine reductase (DHPR) activity, analysis of urinary pterins, and oral tetrahydrobiopterin (BH₄) loading test. Twenty-seven cases (1/31,000) of PKU, including 22 classical PKU, 4 BH₄ synthesis deficient PKU and one DHPR deficient PKU, were confirmed. All of the PKU patients were treated accordingly within 37 days after birth. Except very mild mental development delay in two of the BH₄ synthesis deficient patients, the physical and mental developments of these patients are normal at the present time. From this data, the incidence of BH₄ deficient PKU in Taiwan is estimated to be around 1/168,000, which is much higher than that in Caucasian and Japanese. This result indicates that differential diagnosis of variant forms of PKU is very important for Chinese PKU patients. This integrated nationwide neonatal screening program developed by us provides a very effective health service to prevent mental retardation caused by different forms of PKU in Taiwan.