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9. CLASSICAL AND TETRAHYDROBIOPTERIN DEFICIENT PHENYLKETONURIA IN TAIWAN

Kwang-Jen Hsiao [1,2], Tsung-Sheng Su [1,2], Kuang-Dong Wuu [2].

Dept of Medical Research[1], Veterans General Hospital, and Institute of Genetics[2], National Yang-Ming Medical College; Taipei, Taiwan 11217; Republic of China

To study the morbidity of phenylketonuria (PKU) in mentally retarded children, blood of 551 institutionalized children and 4,744 mentally retarded schoolchildren in Taiwan was collected on filter paper between 1982 and 1983. PKU was screened by determination of phenylalanine by the Guthrie method. Four cases of PKU were found. Urinary pterins analysis by high performance liquid chromatography (HPLC) showed that one case was caused by tetrahydrobiopterin (BH4) synthesis deficiency. The incidence of PKU in mentally retarded children was estimated at around 0.06-0.2%. Therefore, a project to develop a nationwide neonatal screening program for early diagnosis and treatment has been established. From 1984.1 to 1989.3 the capillary blood of 222,800 newborns was collected on filter paper about five days after birth by 249 delivery units all over Taiwan. The dried blood samples were mailed to the central laboratory for screening of blood phenylalanine and other congenital metabolic diseases. Six PKU babies (1/38,000) were detected. There were two cases with defective synthesis of BH4 and one case with dihydropteridine reductase (DHPR) deficiency. All cases of PKU detected by our neonatal screening program were confirmed by BH4 oral loading test, urinary pterins analysis, and blood DHPR determination; appropriate treatment was begun within 37 days of birth.

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