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SCREENING FOR INHERITED METABOLIC DISEASES AND CONGENITAL HYPOTHYROIDISM IN 4,744 MENTALLY RETARDED SCHOOL CHILDREN IN TAIWAN

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Summary For the purpose of exploring the possibility of implementing a nation-wide screening program for inherited metabolic diseases and congenital hypothyroidism in Chinese newborn infants, a pilot study was initiated in 1983 to detect patients with phenylketonuria (PKU), galactosemia, homocystinuria, maple syrup urine disease and congenital hypothyroidism (CHT) in mildly mentally retarded (mostly IQ 50–75) school children in Taiwan. Of 4,744 blood samples collected on filter paper from 246 primary and junior high schools all over the island, preliminary screening disclosed six suspected positive cases of PKU and nine of CHT. Two cases of classical PKU, one case of atypical PKU caused by tetrahydrobiopterin deficiency, and seven cases of CHT (one hypoplastic, two athyroid, three ectopic and one dyshormonogenesis) were finally confirmed. In addition to the seven cases of CHT, one more case found by questionnaire survey. This case was missed from the screening because he was recognized as CHT previously and was on thyroxine replacement therapy during screening. The incidence is 1/1,581 for PKU and 1/593 for CHT in these children.

INTRODUCTION

Although neonatal screening for metabolic diseases has become routine preventive pediatrics in some of the developed countries (Bickel, 1980), no such measure on a nationwide scale has been initiated with the Chinese newborn in Taiwan. Basic information such as the incidence of phenylketonuria or other metabolic diseases is lacking. With the aim of establishing a nationally neonatal screening program

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