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CONGENITAL METABOLIC DISEASES IN MENTALLY RETARDED CHINESE CHILDREN: SCREENING FOR CONGENITAL HYPOTHYROIDISM, PHENYLKETONURIA, GALACTOSEMIA, MAPLE SYRUP URINE DISEASE AND HOMOCYSTINURIA.

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It was believed that phenylketonuria (PKU) is a rare inborn metabolic disease in Chinese population and very few cases have been reported in the past. For studying the morbidity of PKU and other potentially treatable inborn metabolic diseases in Taiwan, the blood of 4744 children were collected on filter paper from a total of 4994 registered students in the public school mentally retarded classes all over Taiwan in the Spring of 1983. Most of those students had IQ between 50 and 80.

The blood phenylalanine (Phe) were screened by the Guthrie's becteria inhibition assay. Three of them were positive (Phe 14-20 mg/dl) and were recalled. Two cases of PKU (male, 16 yr. & female, 11 yr.) and one case of hyperphenylalaninemia (male, 13 yr.) were confirmed by positive urinary $FeCl_3$ test, serum Phe (fluorometric method) of 26.7, 33.1, 13.6 mg/dl and Phe/Tyrosine ratio (amino acid analyzer) of 27.0, 51.0, 13.4, respectively. Two elder sisters, one of each PKU patient, were also diagnosed as PKU with mental retardation in the follow-up family study. Using the same screening and confirmatory methods, a 12 years old PKU boy was detected from 550 institutionized children in northern Taiwan. A 20 months old boy and an 8 years old girl were identified as PKU from 147 selected pediatric neuropsychiatrical patients in our hospital within a year (Nov. 1982-Oct. 1983)

The congenital hypothyroidism (CHT) was screened by the blood thyrotropin (TSH) enzymeimmunoassay. The abnormal results (>10 μU/ml of blood) were found in 9 specimens and were recalled. Seven cases of congenital hypothyroidism (agenesis: 2, dysgenesis: 1, ectopic: 3, dyshormonogenesis: 1?) and a case of Down's syndrome (Tyisomy 21) with autoimmune thyroiditis were confirmed by serum TSH, T4, T3, bone x-ray, thyroid scan, antithyroid antibody and chromosome analysis. In the 551 institutionized children, 5 abnormal TSH results were detected by radioimmunoassay. All of those suspected cases were recalled for confirmatory tests, except one with TSH 55 μU/ml who has not been able to be located. One each of athyroid and ectopic thyroid, and two cases of Down's syndrome (Trisomy 21) with autoimmune thyroiditis were also confirmed.

Except one case of congenital hypothyroidism, who was receiving proper therapy, none of known cases of PKU and CHT was missed by these screening methods. The maple syrup urine disease, homocystinuria and galactosemia were screened by blood leucine (Guthrie's test), methionine (Guthrie's test) and galactose (E. Coli – phage assay), respectively. But none of true positive was found for these three diseases.

From these data, the morbidity of PKU in mentally retarded Chinese children may be estimated at around 0.04-0.2% and may be as high as 1.4% in clinically selected pediatric patients. These results show that PKU does cause mental retardation in Chinese children on this island and may not be as rare as we thought. And the morbidity of CHT in mentally retarded Chinese children may be estimated at around 0.15-0.5%. Therefore, a neonatal screening study is indicated both for estimating the prevalence of these potentially treatable inborn metabolic diseases in Chinese and for early diagnosis and treatment to prevent those affected babies from mental retardation in Taiwan.