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P-38 SCREENING FOR TREATABLE INBORN METABOLIC DISEASES IN MENTAL RETARDED CHINESE CHILDREN

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For studying the morbidity of potentially treatable inborn metabolic diseases in Taiwan, the blood of 551 institutionized children in northern Taiwan were collected on filter paper. The specimens were sent to Hamburg in dry ice and were screened for congenital hypothyroidism (CHT), phenylketonuria (PKU), homocystinuria (HCU), maple syrup urine disease (MSUD) and galactosemia. The abnormal results were found in 5 specimens for TSH and 11 specimens for phenylalanine. There were 2 specimens with slightly elevated galactose and one with slightly elevated leucine. All of those suspected cases were recalled for confirmatory tests, except one with elevated TSH (55 uU/ml) who has not been able to be located. A 12 years old boy was confirmed as PKU. An 8 years old boy and a 19 years old girl were identified as CHT with aplastic and ectopic thyroid, respectively. But not MSUD, HCU nor galactosemia was confirmed. From the result, the morbidity of CHT and PKU in mental retarded Chinese children may be estimated around 0.4—0.5% and 0.2%, respectively. These data indicated that CHT and PKU do cause mental retardation in children on this island and we should look into neonatal screening program for early diagnosis and treatment to prevent mental retardation in those affected babies in Taiwan.