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智能殘障兒童先天性甲腺低能症之篩檢
Screening for Congenital Hypothyroidism in Mental Retarded Children.

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For studying the morbidity of Congenital Hypothyroidism, a potential treatable inborn metabolic disease, in mental retarded children in Taiwan, the blood of 551 institutionized children in northern Taiwan were collected on filter paper (S & S No.2992) in the Spring of 1982. The specimens were sent to Hamburg in dry ice and the blood thyrotropin (TSH) was determined by solid phase radioimmunoassay. Five specimens were found abnormal with TSH higher than 20 uU/ml. Four of them were recalled for confirmatory tests, including serum TSH, Thyroxine (T_4), Triiodothyronine (T_3), bone X-ray and thyroid-scanning with ^{99m}Tc or ^{131}I. One of them with elevated TSH (55 uU/ml blood) has not been able to be located.

An 8 years old boy (Screen TSH > 112 uU/ml blood) and a 19 years old girl (Screen TSH > 50 uU/ml blood) were identified as congenital hypothyroidism with aplastic and ectopic thyroid, respectively. The other two cases were found to be Down's syndrom (21 trisomy) with hypothyroidism. The screening TSH results were 20-35 uU/ml for both of them. The recalled abnormal cases were

also confirmed by dry blood spot TSH enzyme immunoassay performed locally. From this result, the morbidity of congenital hypothyroidism in mental retarded Chinese children may be estimated around 0.4-0.5% (2 - 3/551). A neonatal screening program is indicated for early diagnosis and treatment of this disease to prevent mental retardation in those affected babies in Taiwan.