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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.

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台灣地區嬰幼兒苯酮尿症飲食治療之研究

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苯酮尿症（phenylketonuria，簡寫爲 PKU），係一種先天性代謝異常疾病，病人因缺乏 phenylalamine hydroxylase，使 phenylalamine不能代謝成 tyrosine，致代謝產物堆積體內造成重度智能不足，但若早期診斷在出生兩個月內開始治療，其智力之增長可與正常孩童相當，目前唯一而有效之治療法爲採用低苯丙胺酸含量之飲食控制，飲食之攝取需使血清苯丙胺酸值維持在 2～8 mg/dl，目前接受治療之病童，經一年半之控制顯示智力上有增進，對腦部發育已停止之較年長孩童，則在行爲表現上有顯著改善，然對苯丙胺酸之需量個別差異極大（22.6±3.61/kg ～ 124 ± 25mg/kg），我國目前因接受治療之病人人數太少，且治療時間太短，無法找出我國嬰幼兒對苯丙胺酸需要量之參考範圍，以後如能對新生兒進行全面篩檢，每年約可找到 20 名患者（每年之新生兒約 40 萬，估計每 2 萬人中有一名），則經年累月患者人數亦相當可觀，由長期之治療研究期能建立一適用於我國 PKU 患者之營養素需要量的建議參考值，以做爲日後患者飲食治療原則之一典範。