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應用甲促素酶免疫測定法篩檢中國新生兒中之先天性甲腺低能症

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Screening for Congenital Hypothyroidism in Chinese Neonates by TSH Enzymeimmunoassay

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Early detection and treatment of congenital hypothyroidism (CHT) may prevent a servere sequela--mental retardation. Cretinism is still very frequently found in pediatric clinics all over Taiwan. Our previous study has shown that the morbidity of CHT in mentally retarded Chinese children is estimated at around 0.15-0.5%. Therefore, a pilot neonatal screening study is indicated both for estimating the prevalence of CHT and for developing a nationwide neonatal screening program for early diagnosis and treatment to prevent those affected babies from mental retardation.

During the year of 1984 (1984.1.1-12.31), the blood of 9952 newborns were collected on filter paper by heel skin puncture around five days after birth from nine hospitals, one obstetric clinic, and Nan-Tou county health stations. This represents an effective collection rate of over 97%. The dry blood samples were mailed to the screening center, Clinical Biochemistry Research Laboratory, Veterans General Hospital, Taipei. Thyrotropin (TSH) of those dry blood samples were determined by a double antibody

competitive enzymeimmunoassay (EIA). The antigen, TSH, was labeled with galactosidase, and was detected fluorescently with 4-methylumbelliferyl-galactoside. Abnormal result (>10mU/L of blood) were found in 61 specimens (positive rate: 0.61%) and 51 of them were recalled by the end of January, 1985. Seven out of 51 recalled samples were still positive, but three of them with relatively low blood TSH became negative on second follow up sample. Four cases of CHT, 3 female and 1 male, were confirmed by serum TSH, T_4, T_3, bone X-ray and ^{99m}Tc thyroid scan. There was one case each of athyroid and ectopic thyroid and a possible case of dyshormonogenesis, who is waiting for final re-evaluation at two years of age. The fourth case, who was born in Hualien, is waiting for thyroid scan for final diagnosis. All of the confirmed cases of CHT were diagnosed and put on thyroxine replacement therapy within the first 23 days of their life. They are developing normally at the present time.

From these data, the prevalence of CHT in Taiwan may be estimated at around one in 2500, which is about two times that of North America, Australia, Japan and some European countries. The reason for this relatively high prevalence of CHT in Taiwan remains to be elucidated. These results show that this pilot screening program could provide very effective preventive medical service in Taiwan. With the high prevalence of CHT in Taiwan, a nationwide neonatal screening program based on this model system to prevent those affected babies from mental retardation is highly indicated.

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