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A PILOT STUDY ON NEONATAL SCREENING OF CONGENITAL ADRENAL HYPERPLASIA IN TAIWAN

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Congenital adrenal hyperplasia (CAH) is a family of inherited disorders of adrenal steroidogenesis, caused most commonly by 21-hydroxylase deficiency. Enzymeimmunoassay (EIA) kits (kit C: CIBA-Corning Diagnostic; kit E: EIKEN Chemical) for determination of 17-hydroxyporgesterone (17-OHP) in dried blood spot collected on filter paper were evaluated by our laboratory for screening of congenital adrenal hyperplasia. Direct analysis of 17-OHP in dried blood spot was found to be interfered by water-soluble steroids which may cause false positive screening results. Method for extracting blood samples with diethyl ether was established to improve the specificity. The precision, analytical recovery and linearity of both direct and extraction methods were satisfactory. Reference range of direct and extraction methods were estimated to be 4.4-31.0 ng/ml(blood) and 1.4-7.8 ng/ml for kit C, and 1.0-8.0 ng/ml and 0.6-5.5 ng/ml for kit E, respectively, in normal Chinese newborns. High blood 17-OHP values (>80 ng/ml) were found by both methods for the known CAH cases (3 children and a newborn at age of 6 days). 17-OHP of low birth weight newborns determined by direct method was significantly higher than those of normal birth weight newborns, but no significant difference was found when 17-OHP was analyzed by extraction method. The result indicated that the extraction method will reduce the false positive screening results and could be used as a preliminary back-up method for the simple direct EIA screening test. For fast confirmatory diagnosis of CAH screening positive cases, a reversed phase (C-18) high performance liquid chromatography system with UV (250nm) detection for simultaneous analysis of several serum steroids was established.

From 1989.5 to 1990.4, 16,023 Chinese newborns in Taiwan were screened with kit C. 364(2.3%) of them were found to be positive (>30 ng/ml) by direct method. From these positive cases, only 57(15.7%) of them had 17-OHP >6 ng/ml when tested by the extraction method. During 1991, 20,470 newborns were screened with kit E, 339(1.7%) of them were found to be positive(>8 ng/ml) by direct method. 107(31.6%) of these positive cases were still positive (>6 ng/ml) with extraction. Except 5 cases lost in the follow-up, all of the screening positive cases detected by extraction were recalled, but no CAH case was confirmed. The data indicated that the EIA methods and the system developed in this study may be suitable for mass neonatal screening of CAH in Taiwan, but the incidence of CAH in Chinese remains to be determined.

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