Colloquim on Inborn Errors of Metablism (Med Res Council, South Africa and National Science Council, R.O.C.), 1989;p.11-12.

'RICA

'isorders, Department of
Town, Medical School,

'equently in the various
minent among these are
Afrikaner community,
ınd Tay-Sachs disease in
osphate dehydrogenase
ıssaemia in the Greek
:inosis and cleidocranial
try, diabetes mellitus in
tosa in the Tristan da
syndromes have been

ıotable by their virtual
ısis and phenylketonuria

f genetic conditions in a
: practical importance
re disease and in the

3. NEONATAL SCREENING FOR INBORN METABOLIC DISEASES IN TAIWAN

Kwang-Jen Hsiao [1,2] and Kuang-Dong Wuu [2]

Dept. of Medical Research[1], Veterans General Hospital, and Institute of Genetics[2], National Yang-Ming Medical College; Taipei, Taiwan 11217; Republic of China.

In order to study the incidence of potentially treatable congenital metabolic diseases in mentally retarded children and to establish screening tests, diagnostic procedures and therapeutic methods, blood of 551 institutionalized children and 4 744 mentally retarded school-children in Taiwan was collected on filter paper between 1982 and 1983. Congenital hypothyroidism (CHT), phenylketonuria (PKU), galactosemia, maple syrup urine disease (MSUD), and homocystinuria were screened by determination of TSH (EIA), phenylalanine (Guthrie), galactose (E.Coli-Phage) and transferase (Beutler), leucine (Guthrie), and methionine (Guthrie) on the dried blood spots. Four cases of PKU and 10 cases of CHT were detected. The incidence of PKU and CHT in mentally retarded Chinese children was estimated at 0.06-0.2% and 0.15-0.5% respectively. Therefore we developed a pilot nationwide neonatal screening program for early diagnosis and treatment. From 1984.1 to 1985.7 capillary blood of 18 316 newborns was collected on filter paper, about five days after birth, by 13 hospitals, Nan-Tou County health stations, and obstetric clinics and mid-wives in Hsin-Chu County. The collecting system included all types of delivery on the island. The effective collection rate was 95%. The dry blood samples were mailed to the screening centre and screened to the five congenital metabolic diseases. Cases with high positive values were immediately referred to one of the 4 local diagnosis and treatment centres. In borderline positive cases a 2nd sample was collected. Where the result was positive, the case was referred to a local centre. Eight cases of CHT, one of PKU and one of glactosemia were confirmed. All cases of CHT and galactosemia were diagnosed and treated within 23 days of birth. In the case of PKU, which is caused by tetrahydrobiopterin synthesis deficiency, treatment was commenced by the 37th day of life. All patients are developing normally at present.

(108)