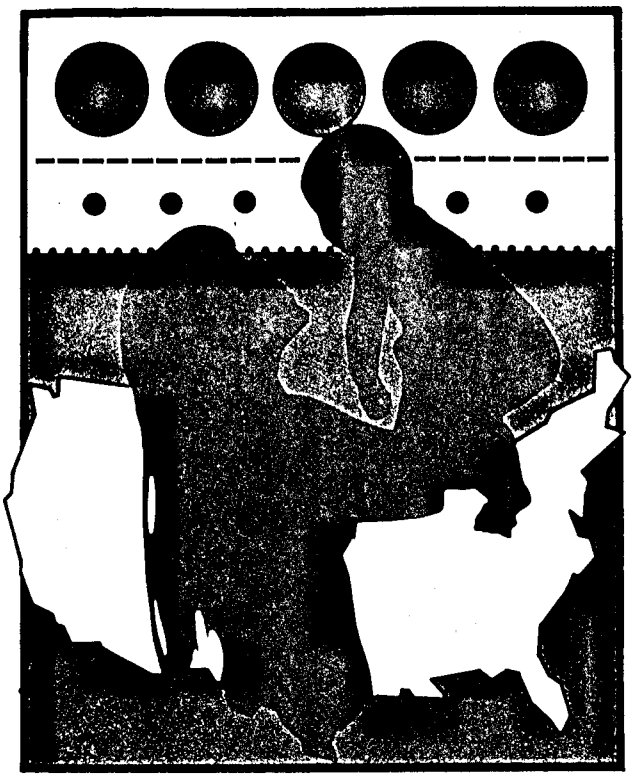

NEONATAL SCREENING

PROCEEDINGS OF THE 6th NATIONAL NEONATAL SCREENING SYMPOSIUM

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NEONATAL SCREENING FOR GLUCOSE-6-PHOSPHATE DEHYDROGENASE (G6PD) DEFICIENCY IN TAIWAN. Kwang-Jen Hsiao*, Mei-Hsueh Chen, Szu-Hu Chaing. Clinical Biochem. Res. Laboratory, Dept. of Medical Research, Veterans General Hospital, Taipei, Taiwan 11217, Republic of China.

G6PD deficiency is one of the most common enzyme deficiency of human beings. A pilot project to establish a nationwide neonatal G6PD screening program in Taiwan was started in 1984. Heel or cord blood collected on filter paper was screened by a qualitative fluorometric method. The positive samples was re-checked by a semi-quantitative fluorometric method which was developed in our laboratory. The final positive cases were referred to one of the six local centers for confirmatory tests, medical care and genetic counseling. From 1984.11.1 to 1987.6.30, 82,653 samples were collected from all over Taiwan. 2,610 cases (3.2%) were found to be positive by screening. Among 1,335 successfully recalled cases, 890 were confirmed as G6PD deficiency. No kernicterus has been reported in the G6PD deficient newborns detected by this program. Based on these results, the G6PD screening has been incorperated into the national neonatal screening program in Taiwan since July, 1987.

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LONG-TERM MANAGEMENT OF PHENYLKETONURIA IN THE STATE OF WASHINGTON. Sheila Neier*, Roberta Spiro, Horace C. Thuline. Division of Public Health, DSHS, State of Washington, Seattle, Washington.

A comprehensive approach exists for all elements of newborn screening for metabolic disease in the State of Washington. Following identification by the laboratory tests and clinical assessment for dietary management of PKU, all patients enter the interdisciplinary PKU clinic supported by the State Genetics Services Section. This clinic is at the University of Washington Child Development and Mental Retardation Center. The staff at the PKU clinic provide the children and parents with lifetime dietary modification skills. Currently, of the 66 children, adolescents and young adults being followed at the PKU clinic, 88% are seen regularly for clinic appointments. Dietary compliance data in 1986 show 70% of the children with excellent serum phenylalanine levels (2 - 10 mg%) and an additional 21% in moderately effective control (11 - 15 mg%). Special efforts are being made to improve dietary control during the difficult years of adolescence, especially for sexually active females.

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TEN YEARS OF MANDATED NEWBORN SCREENING IN WASHINGTON.
H.C.Thuline*, Michael W. Glass, Sheila Neier, Roberta Spiro.
Division of Public Health, DSHS, State of Washington, Seattle, WA

In 1976 the state legislature mandated screening of newborns for PKU and other metabolic disorders leading to mental retardation or handicaps before the infants are discharged from the hospital. By October 1977 the program to screen for PKU and congenital hypothyroidism (CHT) was in place. Screening for congenital adrenal hyperplasia (CAH) was added in July 1984 as a research project then became part of the routine screening in July 1987. Over 630,000 infants have been screened for PKU and CHT. CAH screening has been done for 228,000 of these. Program elements beyond screening and notification of physicians have been developed and added. These include follow-up for children with CHT, funding of the PKU clinic, stocking of dietary products at a cost-saving to families, and follow-up for maternal PKU. The full program will be described in more detail.