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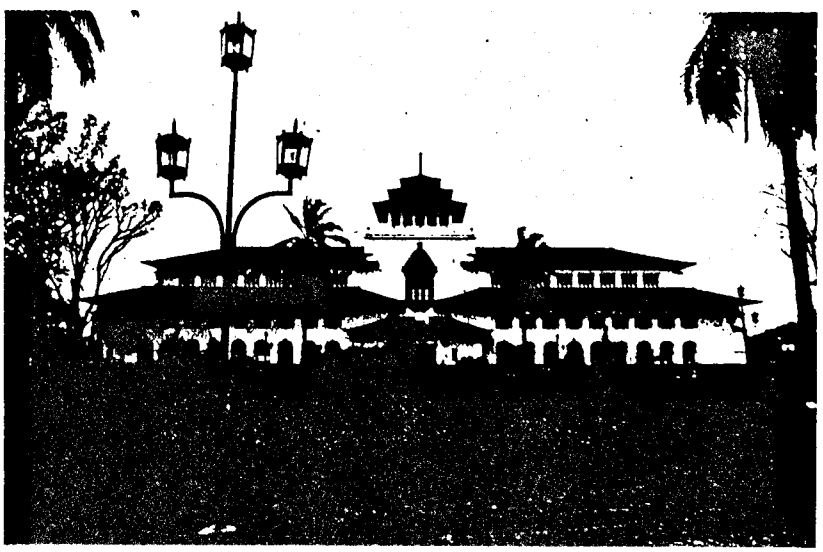

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NEONATAL SCREENING STRATEGY FOR CONGENITAL METABOLIC DISEASES

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Some of the congenital metabolic disorders have no clinical symptoms during neonatal period, if not treated early irreversible damages such as mental retardation will occur. The permanent damages can be avoided if these diseases are able to be detected biochemically by neonatal screening in the early stage of life, and treated immediately with appropriate therapy.

According to the treatable and detectable criteria, diseases such as phenylketonuria (PKU), maple syrup urine disease, galactosemia, homocystinuria, congenital adrenal hyperplasia, congenital hypothyroidism (CHT) are suggested to be included in neonatal screening programs. For high risk population of glucose- 6-phosphate dehydrogenase (G6PD) deficiency, routine screening program, the blood collected on filter paper, which needs about 0.25 ml heel blood and can be sent by mail, is the method of sample collection used worldwide. Between 1984-1991, 579,572 newborns in Taiwan were screened by our laboratory. G6PD deficiency (2.0%), CHT (1/2,100) and PKU (1/32,000) were commonly detected. System for follow-up positive result is important to prevent irreversible damage to the patients. The screening positive case must be recalled in time to be differentially diagnosed and proper therapy started as early as possible. Continuous monitoring of therapy should be prosecuted to ensure that the patients are in well development. Indeed, neonatal screening requires a network with the laboratory staff, nurse coordinators, pediatricians, dietlcians, and other related health workers fully involved for a successful prevention program.

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