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GENETIC DISORDERS AND NEONATAL SCREENING

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Mass screening of inborn errors of metabolism (IEM) at newborn stage is proved to be an effective prevention program for some mendelian genetic disorders. Some of the congenital 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 in the early stage of life, and treated immediately with appropriate therapy. For regional centralized neonatal screening program, the blood collected on filter paper, which needs about 0.3ml heel blood and can be sent by mail, is the method of sample collection used worldwide.

According to the treatable and detectable criteria, diseases such as phenylketonuria (PKU), maple syrup urine disease(MSUD), homocystinuria, galactosemia, congenital adrenal hyperplasia are suggested to be included in neonatal screening programs. For high risk populations of glucose-6-phosphate dehydrogenase deficiency, routine screening are also considered. With respect to its high incidence(about 1:4000) and to the ease and efficiency of its treatment, congenital hypothyroidism (CHT) is also recommended to be screened, although 80% CHT are not caused by genetic defects. The benefit/cost ratio for neonatal screening of PKU and CHT were estimated to be 1.7 and 2.0, respectively, in Taiwan during 1988-1989.

System for follow-up positive screening results 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 the patients are in well development. Indeed, neonatal screening requires a network with the laboratory staff, the nurse coordinators, pediatricians, dieticians, and other related health workers fully involved for a successful prevention program.

Diseases which are not treatable at present time are not recommended to be included in neonatal screening for genetic counseling only. However, currently prophylactic therapy has been tried for sickle cell anaemia, once it has been proved to be effective, the disease will be considered for high risk population screening. The genes of Duchenne's muscular dystrophy (DMD) and cystic fibrosis had been cloned recently, and its pathophysiology are in heavy study. These diseases should be considered to be included in neonatal screening program as soon as the effective therapies are developed.

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