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遺傳醫學的現在與未來

The Present and Future of Medical Genetics

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Developments of Neonatal Screening in Taiwan
(台灣地區新生兒篩檢之回顧與展望)

Kwang-Jen Hsiao (蕭廣仁)[1,2] and Szu-Hui Chiang (蔣思慧)[2]

Institute of Genetics and Genome Research Center[1], National Yang Ming University, University System of Taiwan, and Department of Medical Research and Education[2], Taipei Veterans General Hospital; Taipei, Taiwan 112, R.O.C.

Some of the congenital metabolic disorders have no specific 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 are able to be detected biochemically by neonatal screening in the early stage of life, and treated immediately with appropriate therapy and intervention. Method development pilot programs (including dried blood sample collecting, screening tests, confirmatory diagnostic procedures and treatments) were carried out in mental retarded children between 1982 and 1983 in Taiwan. Based on the methods developed, the nationwide project to set up neonatal screening for congenital hypothyroidism (CHT), phenylketonuria (PKU), maple syrup urine disease (MSUD), homocystinuria (HCU), and galactosemia (GAL) was started in January 1984. After the nationwide neonatal screening system was established in July 1985, the method for screening of glucose-6-phosphate dehydrogenase (G6PD) deficiency was developed. The incidence of G6PD deficiency was estimated to be about 2% (male 3%, female 0.9%) in Taiwan based on the screening program. Since no MSUD was found from 200,000 newborns screened, after a two year (1985.7-1986.7) pilot study on G6PD screening, MSUD was replaced by G6PD in the routine nationwide neonatal screening program. The screening coverage rate in Taiwan has reached 80% in 1990 and 99% since 1996. From 1984.1 to 2002.12, 4,462,600 newborns have been screened. The incidences of CHT, PKU, HCU, and GAL were reported to be about 1/1,800, 1/31,400, 1/263,000, and 1/744,000, respectively. Most of the affected cases were detected and treated accordingly within one month of birth and are developing normally at the present time.

Each of the three neonatal screening centers in Taiwan has started individual voluntary program paid by the parents for selective screening of congenital adrenal hyperplasia (CAH) and defects in other amino acids and acylcarnitines metabolisms, which were detected by tandem mass spectrometry (MS/MS), since 2000.3 and 2001.8, respectively. The incidence of CAH was estimated around 1/15,000 from 600,000 newborns screened between 2000.3 and 2003.10. Six cases of 3-methylcrotonyl-CoA carboxylase deficiency (3MCC), 4 cases of citrullinemia (CIT), 3 cases of MSUD, 2 cases of glutaric aciduria type I (GAI), 1 case of methylmalonic aciduria (MMA), and 1 case of nonketotic hyperglycinemia were detected from 216,000 newborns by the MS/MS screening between 2001.8 and 2003.10.

Recently, a technological assessment research supported by the Bureau of Health Promotion, Department of Health has reached a consent recommendation about the adjustment of the items for neonatal screening in Taiwan: 1). the 5 current routine items should be kept, 2). CAH, MSUD, MCAD, CIT, GAI, MMA, and isovaleric academia (IVA) should be included as routine items, 3). biotinidase deficiency, argininosuccinase deficiency, propionic academia, and C5OH-carnitine should be included as routine items for a pilot project, 4). any other disease which could be detected by MS/MS should be considered as a research item only at the present time, 5). any disease incorporated into the routine services, including pilot project items, should have confirmatory diagnosis and follow up treatment system prepared in place before its screening program starts, 6). the positive results of CAH, G6PD, GAL, and MS/MS tests should be referred for follow-up no later than 7 days after birth, 7). the routine screening items should be available to all the newborns non-selectively, 8). the routine screening items should be reviewed periodically.