The experience of neonatal screening in Taiwan

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A nationwide program for neonatal screening of congenital metabolic diseases in Taiwan was started in 1984. There were two screening centers, one located at Veterans General Hospital-Taipei (VGH) and the other located at National Taiwan University Hospital. From 1984.1 to 1993.9, 838.572 newborns were screened for phenylketonuria (PKU), homocystinuria, maple syrup urine disease (MSUD), galactosemia, congenital and hypothyroidism (CHT) by the VGH screening center. Dried blood spot samples collected in filter paper by heel puncture were sent to VGH screening center from 445 delivery units, including 111 hospitals, 260 obstetric clinics, 23 midwives and 51 health stations, all over Taiwan by mail. One cases of homocystinuria, two cases of galactosemia, 27 cases of PKU (1/31.000, including 22 classical PKU, 4 BH_4 synthesis deficient and one DHPR deficient PKU), and 401 cases of CHT/1/2.100), were confirmed. Most of them were diagnosed and treated within 3 weeks of life. The mass neonatal screening of glucose-6-phosphate dehydrogenase (G6PD) deficiency by flourometric spot test in Taiwan was started with educational program in 1984. Seventeen G6PD referral centers connected by fax were established in local hospitals island wide to follow up positive cases with confirmatory tests, medical care and genetic counseling. Since no MSUD was found in 220.000 neonates screened in Taiwan, after a two-year (1985.7 – 1987.6) pilot study on G6PD screening, the MSUD was replaced by G6PD in the routine neonatal screening program in July, 1987. From 1984.11 to 1993.9, 828.509 newborns were screened for G6PD deficiency by the VGH screening center. The incidence of G6PD deficiency is estimated to be around 2.1% (male 3.1%, female 0.9%). The neonatal screening coverage rate in Taiwan has reached to 97.8% (~ 310.000/year) in 1994.