

Workshop

Workshop 3-4

Neonatal screening of glucose-6-phosphate dehydrogenase deficiency

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Glucose-6-phosphate dehydrogenase (G6PD; EC 1.1.1.49) deficiency is the most common enzymopathy in humans. This X-linked genetic disorder (MIM 305900) has been found to be an important cause of neonatal jaundice and acute hemolytic anemia, if some inducing agents (*e.g.* naphthalene, fava bean) are not avoided, in the Southeast Asia and Middle East populations. In order to reduce the complications of G6PD deficiency, such as kernicterus, permanent neurological damage, and death, several populationwide G6PD neonatal screening programs in the Asian Pacific region were started between 1964 (Singapore) and 1980's (*e.g.* Hong Kong, Taiwan). Some Mediterranean (*e.g.* Greece, Cyprus, Lebanon, Turkey) and Southeast Asian countries (*e.g.* Philippines, Thailand, Vietnam) also included the G6PD test in their national or regional neonatal screening program. A few European (*e.g.* Germany, Italy) and American (*e.g.* Washington D.C., Missouri, Pennsylvania) neonatal screening centers have included G6PD test for the selective high risk populations in their region. An external quality assurance (QA) program for the screening test of G6PD deficiency was developed in 1999 in order to assess the reliability of the screening tests. Twelve screening centers (3 in Taiwan, 4 in Mainland China, and one each in Philippines, Thailand, Lebanon, Vietnam, and Turkey) are participating in this QA program at the present time. Recently, most of the neonatal screening centers participating in this QA program have changed to quantitative G6PD screening tests from the qualitative screening test (*e.g.* fluorescence spot test). Two more neonatal screening centers (Mainland China and Philippines) are planning to join this QA program next January (2007). Most of the G6PD mutant alleles (>90%) found in the Southeast Asian screening programs can be detected by analyzing the restriction fragments of the DNA products directly amplified from the dried blood spot by PCR, which can be used to facilitate the confirmatory diagnosis of those screening positive cases. At present, the effective collection rate reached 99.7% of all newborns and the overall incidence rate of G6PD deficiency in Taiwan was about 2%. The exchange blood transfusion and permanent complications caused by G6PD deficiency were dramatically reduced at the present time compared to the period prior to the screening program in Singapore, Hong Kong and Taiwan. The results indicated that neonatal screening could prevent sequela (*e.g.* kernicterus, mental retardation) caused by G6PD deficiency and the G6PD test should be included in the routine neonatal screening programs for the high risk populations.