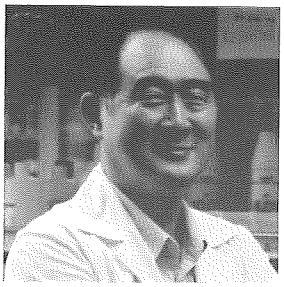
Newborn Screening Society of the Philippines, Inc.

Abstract

NEONATAL SCREENING OF GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY IN ASIA

Kwang-Jen Hsiao, PhD

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. Serveral 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) 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), American (e.g. Washington D.C.), and Australian neonatal screening centers have include G6PD test for the selective high risk populations in their regions. An external quality assurance (EQA) program for the screening test of G6PD deficiency was developed in 1999 in order to assess the reliability of the screening tests. Thirty-five screening laboratories from Taiwan, Mainland China, Philippines, Thailand, Lebanon, Vietnam, Turkey, Germany, Australia, India, Mexico, and Greece, and three reagent manufactures are participating in this EQA program at the present time. Recently, most of the neonatal screening centers participating in this EQA program have changed to quantitative G6PD screening tests from the qualitative ones. Most of the G6PD mutant alleles (>95%) found in the Southeast Asian screening programs can be detected by analyzing 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. Several different common G6PD mutations have been identified in Southern China (1376G>T, 1388G>A, 95A>G), Southeast Asia (383C>T, 487G>A, 871G>A, 1003G>A, 1360C>T), India (131C>G, 563C>T, 949G>A), and Middle East (563C>T, 1003G>A). At present in Taiwan, the effective collection rate has reached 99.7% of all newborns and the overall incidence rate of G6PD deficiency 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.



PROFESSOR KWANG-JEN HSIAO got his Ph.D. in Biomedical Sciences at Mount Sinai School of Medicine, City University of New York, U.S.A.

He is currently Adjunct Research Investigator at the Department of Medical Research & Education, Taipei Veterans General Hospital; Consultant at Taipei City Hospital, Taiwan; Consultant of Taipei Institute of Pathology; and Chief Executive Officer, Preventive Medicine Foundation, Taipei, Taiwan. He was also Vice Chancellor of University System of Taiwan from 2003 to 2004; Professor and Chairman of the Institute of Genetics, National Yang-Ming University, Taiwan; Professor of the Institutes of Biochemistry, Bioinformatics, and Biotechnology in Medicine, and Director of the Genome Research Center of the same University. He has initiated the national newborn screening program in Taiwan between 1982 and 1993.

During his career he won a number of awards including Outstanding Research Scientist, National Science Council, Republic of China; Research Award (Class A), National Science Council, Republic of China; Outstanding Educational Personnel, Ministry of Education, Republic of China; and Health Medal of the Second Order, Department of Health, Republic of China for developing the national newborn screening system.

His current research activities include quality assurance of G6PD activity determination for neonatal screening of G6PD deficiency and developing newborn hearing screening system.

K.-J. Hsiao

10th Newborn Screening Convention

Moving on: Expanded Newborn Screening for Filipino Newborns

October 1 & 2, 2012 SMX Convention Center, Mall of Asia Complex, Pasay City

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