SYMPOSIUM 3

"GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY"

L13 NEONATAL SCREENING OF GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY IN TAIWAN

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Glucose-6-Phosphate dehydrogenase (G6PD) deficiency is the most common enzymopathy of human beings. This X- Linked genetic disorder is a very important cause of neonatal jaundice and acute hemolytic anemia in our region (Southeast Asia and some Pacific islands). The mass neonatal screening of G6PD deficiency by fluorometric spot test in Taiwan was started with education program in 1984. Seventeen G6PD referral centers were established in local hospitals island wide to follow up positive cases with confirmatory tests, medical care and genetic counseling. From Nov.1984 to Sept.1993, 828,509 heel blood samples collected on filter paper from 445 delivery units were screened by the Veterans General Hospital screening center. 27,643 cases with positive result were referred to referral centers by fax immediately after screening. About 77.5% of the positive cases had whole blood G6PD quantitative test and 62.5% of them were confirmed as G6PD deficiency. The incidence of G6PD deficiency is estimated to be around 2.1% (male 3.1%, female 0.9%) in Taiwan. The neonatal screening coverage rate was 96.7% in 1993. In order to facilitate the confirmatory diagnosis process, a method to detect those common southern Chinese G6PD mutations (G1388A, G1376T, C1024T, A493G, G487A, G392T, A95G) was developed by analyzing the restriction fragments of the DNA products directly amplified from the dried blood spot samples by PCR. This analysis detected about 80-90% of G6PD mutant alleles in Taiwan and has been found with similar effectiveness in Canton, Hong Kong, and Singapore. The development of neonatal screening of G6PD deficiency in Taiwan could be used as an example for other areas with high incidence of G6PD deficiency.