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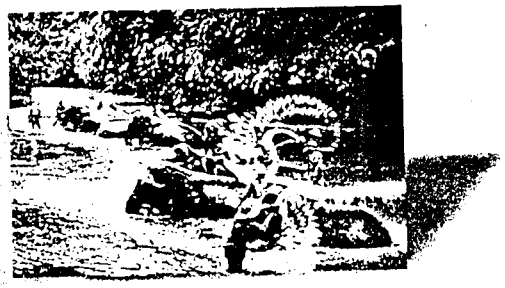
NEONATAL SCREENING IN THE 21ST CENTURY



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Program & Abstracts

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G6PD DEFICIENCY SCREENING IN TAIWAN

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Glucose-6-phosphate dehydrogenase (G6PD) deficiency is the most common enzymopathic disease in Taiwan. The mass neonatal screening of G6PD deficiency by fluorometric spot test in Taiwan was started with a pilot program in 1984. The nationwide screening was started on July 1, 1987, and a follow-up system comprising of eighteen referral hospitals, including outlying islands, was organized for confirmatory test, medical care and genetic counseling. From 1987 to 1997, 2,971,192 heel blood samples collected on filter paper from 1,143 delivery units were screened by four neonatal screening centers. 46,570 cases were confirmed as G6PD deficiency by referral hospitals. 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 99% in 1997. To assess the reliability of the confirmatory test, an external quality assurance (QA) program for G6PD assay was developed. Periodically, 3 or 5 lyophilized quality control materials of different activities of G6PD kept in dry ice were sent to each of the referral hospitals by speed post delivery. The external quality assurance results were evaluated and compared to the reference values determined by our laboratory. From January 1988 to June 1998, 85 quality assurance services were performed. 207 (13.5%) abnormal QA results were found, which were attributed to clerk (24/207, 11.6%), experimental (40/207, 19.3%), and instrumental errors (93/207, 44.9%). 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. The experience of neonatal G6PD screening in Taiwan could be used as an example for other areas with high incidence of G6PD deficiency.