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23 DETECTION OF COMMON GLUCOSE-6-PHOSPHATE DEHYDROGENASE MUTATIONS IN SOUTHERN CHINESE BY POLYMERASE CHAIN REACTION (PCR) USING DRIED BLOOD SPOT SPECIMENS

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Since 1984, dried blood spot collected on filter paper have been used for neonatal screening of glucose-6-phosphate dehydrogenase (G6PD) deficiency and other congenital metabolic diseases in Taiwan. A high incidence (~2%) of G6PD deficiency is found. Recently, the mutations of nucleotide 1388 (G→A), 1376 (G→T), 1024 (C→T), 493 (A→G), 487 (G→A), and 95(A→G) have been reported to account for approximate 80% of G6PD mutant alleles in southern Chinese. In order to reduce recall works of the local G6PD referral hospitals and to facilitate the confirmatory diagnosis process, a method to detect those common G6PD mutations was developed by analyzing the restriction fragments of the DNA products directly amplified from the dried blood spot samples by PCR with mismatched primers. All the primers were designed to include an additional restriction site in the PCR products besides the mutation site for internal quality control. 174 neonatal blood spot samples of confirmed G6PD deficient male patients were collected and analyzed. Among these 174 samples, 141 (81%) were found to have one of the common G6PD mutations. Thirty "1388" (17.2%), seventy-seven "1376" (44.3%), nine "1024" (5.2%), eleven "493" (6.3%), and fourteen "95" (8.0%) were identified. The mutation frequencies are in good agreement with other studies reported in Taiwan. This easy and non-radioactive method provides a way using the same screening dried blood spot specimens to confirm the positive results of the neonatal G6PD screening test, which will alleviate the time and money spent on recall and confirmatory diagnosis. This method could also be applied to detect heterozygotes of G6PD deficiency for genetic counseling, and provides a way to collect samples for large scale epidemiological study of the G6PD mutations in different populations.

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