MUTATIONS IN THE HUMAN GENOME
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TITLE

ANALYSIS OF COMMON GLUCOSE-6-PHOSPHATE DEHYDROGENASE MUTATIONS IN CHINESE BY POLYMERASE CHAIN REACTION USING DRIED BLOOD SPOTS COLLECTED ON FILTER PAPER

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TEXT

Glucose-6-phosphate dehydrogenase (G6PD) deficiency is the most common known hemolytic disease caused by enzyme defect. 400 million people worldwide was estimated to be affected by G6PD deficiency. There is a high frequency (~2 %) of G6PD deficiency in the Chinese population in Taiwan. Six mutations, namely nucleotide 95 A→G, 487 G→A, 493 A→G, 1024 C→T, 1376 G→A, and 1388 G→A, were reported to account for most G6PD mutant alleles in Chinese. Polymerase chain reaction (PCR) was utilized in this study to amplify DNA from blood sample collected on filter paper used in neonatal screening for the easiness of transportation and storage. Dried blood spots specimens from 108 G6PD deficient patients detected by neonatal screening program were collected. The six common G6PD mutations were analyzed by PCR amplification with mismatch primers followed by restriction enzyme digestion. Among the 108 G6PD patients, forty-two "1376"(38.9%), sixteen "1388"(14.8%), eight "493"(7.4%), five "95"(4.6%) and six "1024"(5.6%) mutations were identified. This easy and non-radioactive method provides a way to confirm the positive G6PD deficient cases detected by the neonatal screening test using the same screening dried blood spots specimens. The method could also be applied to detect heterozygotes of G6PD deficiency which were difficult to detect by enzyme analysis. In addition, this method provides a way to collect samples for large scale epidemiological study of the G6PD mutations in different populations.

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